



Genome data

Siv Fjellkårstad
Senior advisor
The Directorate of Health

Today's lecture

1. Introduction
to Genetics

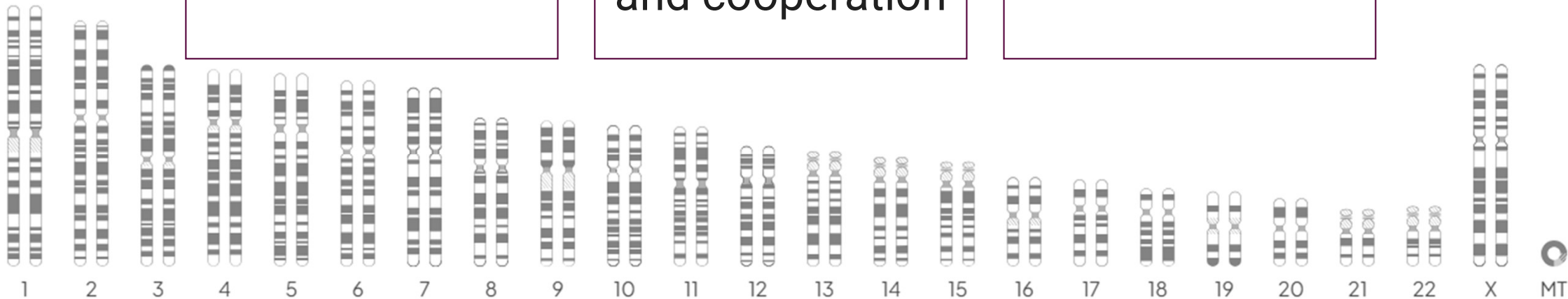
2. Sequencing
and Genomic
Data

3. Genetic Errors
and Conditions

4. Medical
Genetics

5. IT,
infrastructure
and cooperation

6. Ethical
Considerations

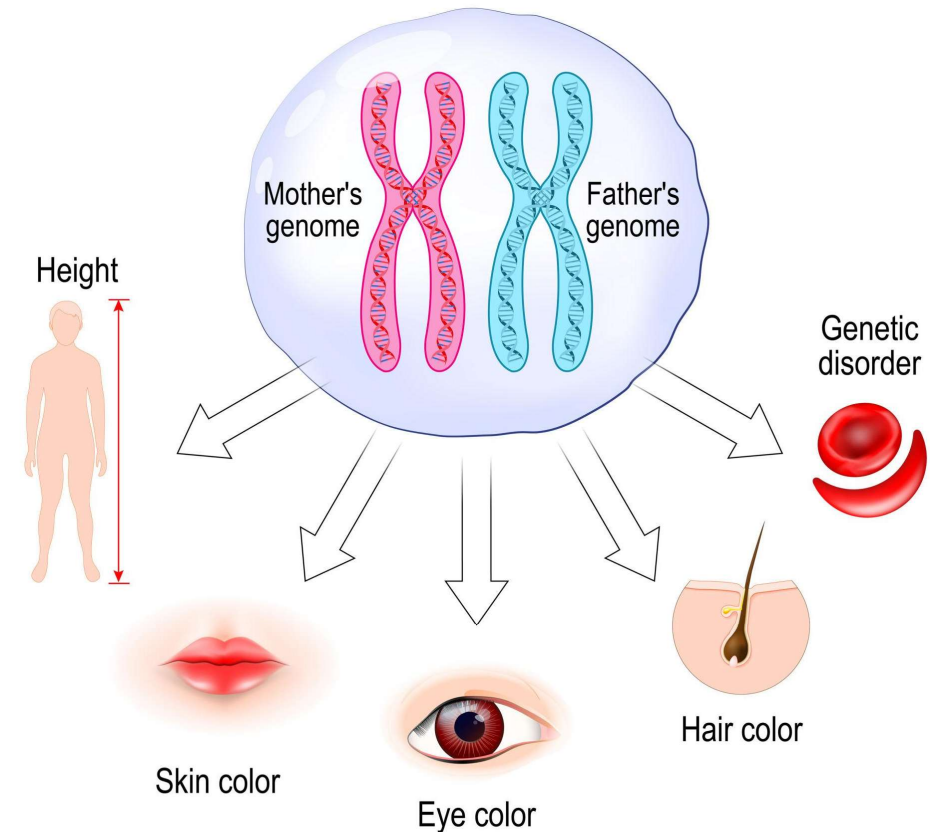


What is genetics?

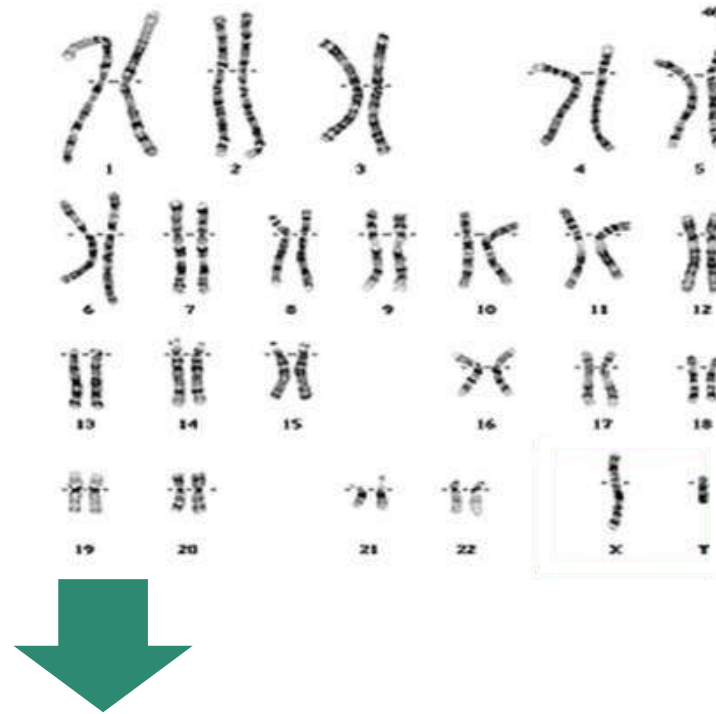
Study of how genetic information is...

- stored in the cell
- expressed
- heriated from parents to offspring

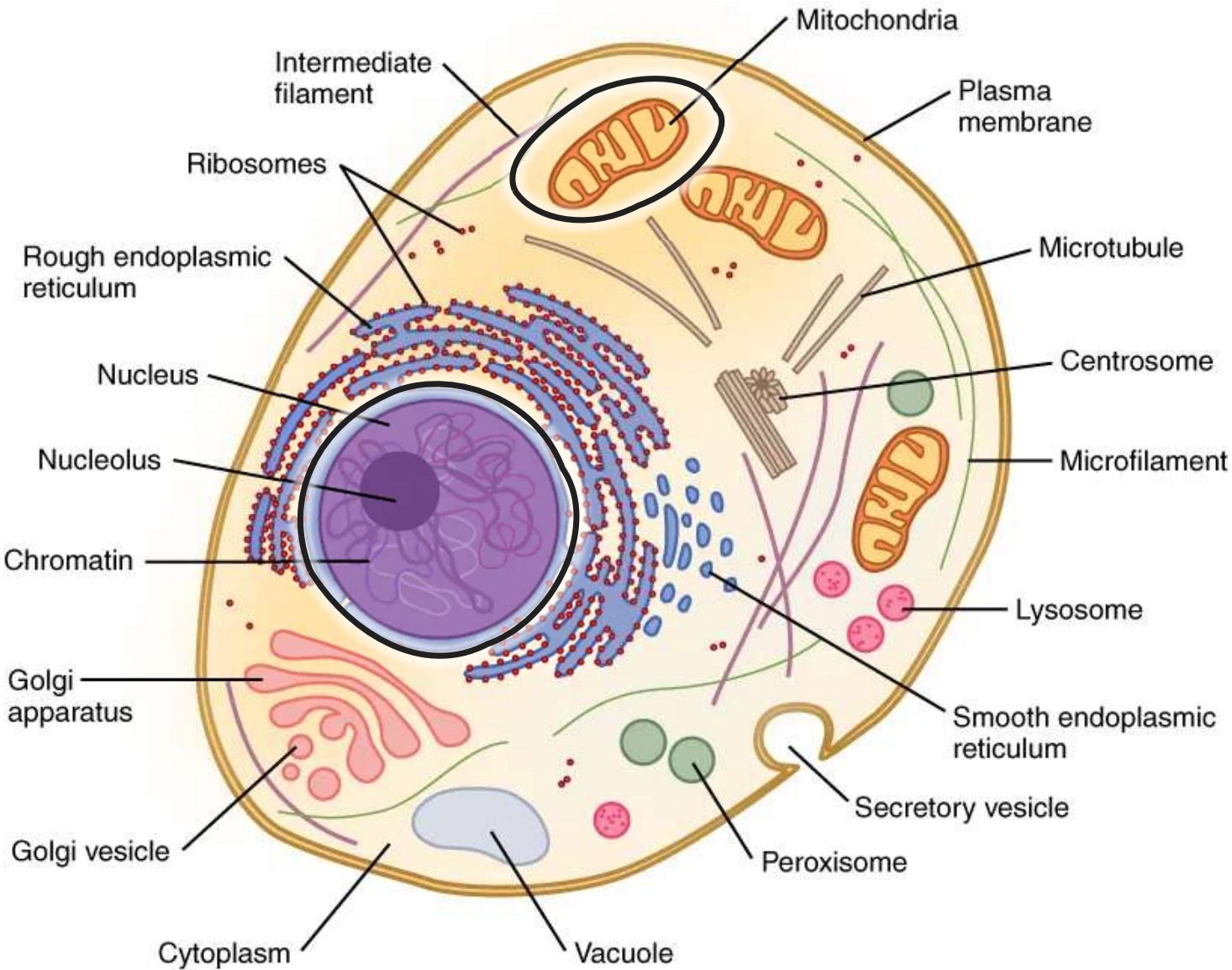
And how it contributes to variation within and between organisms



From DNA to genetic data

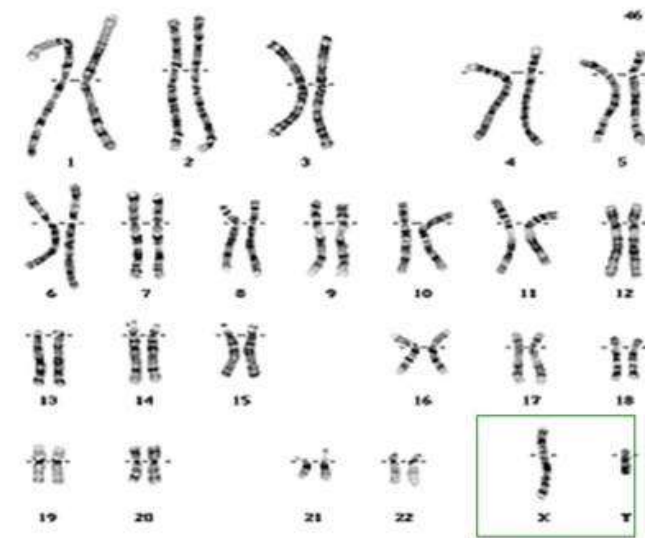


Chr ↕	Position ↕	Variant ID	Gene ↕	Impact	Risk Frequency ↕	Ref	Alt	Your Data ⇅	Risk Version	Your Status ⇅	Condition ⇅	Classification ⇅
1	119422246	rs80358217	HSD3B2	p.Arg249Ter	0.0000948	C	T	CC	T	Harmless (NI)	3 Beta-Hydroxysteroid Dehydrogenase Deficiency ⓘ	Pathogenic
1	119422184–119422211	rs2526392619	HSD3B2	p.Trp230_Ala238del		I	D	II	D	Harmless (NI)	3 Beta-Hydroxysteroid Dehydrogenase Deficiency ⓘ	Pathogenic
1	119422013	rs80358216	HSD3B2	p.Trp171Ter	<0.00001	G	A	GG	A	Harmless (NI)	3 Beta-Hydroxysteroid Dehydrogenase Deficiency ⓘ	Pathogenic
1	119415428	rs765335418	HSD3B2	p.Trp3Ter		G	A	GG	A	Harmless (NI)	3 Beta-Hydroxysteroid Dehydrogenase Deficiency ⓘ	Pathogenic



Human DNA

23 chromosome pairs
in the cell nucleus



Mitochondrial DNA (mtDNA)



Figure: Diagram of typical generalised human cell ([Wikimedia Commons](#))

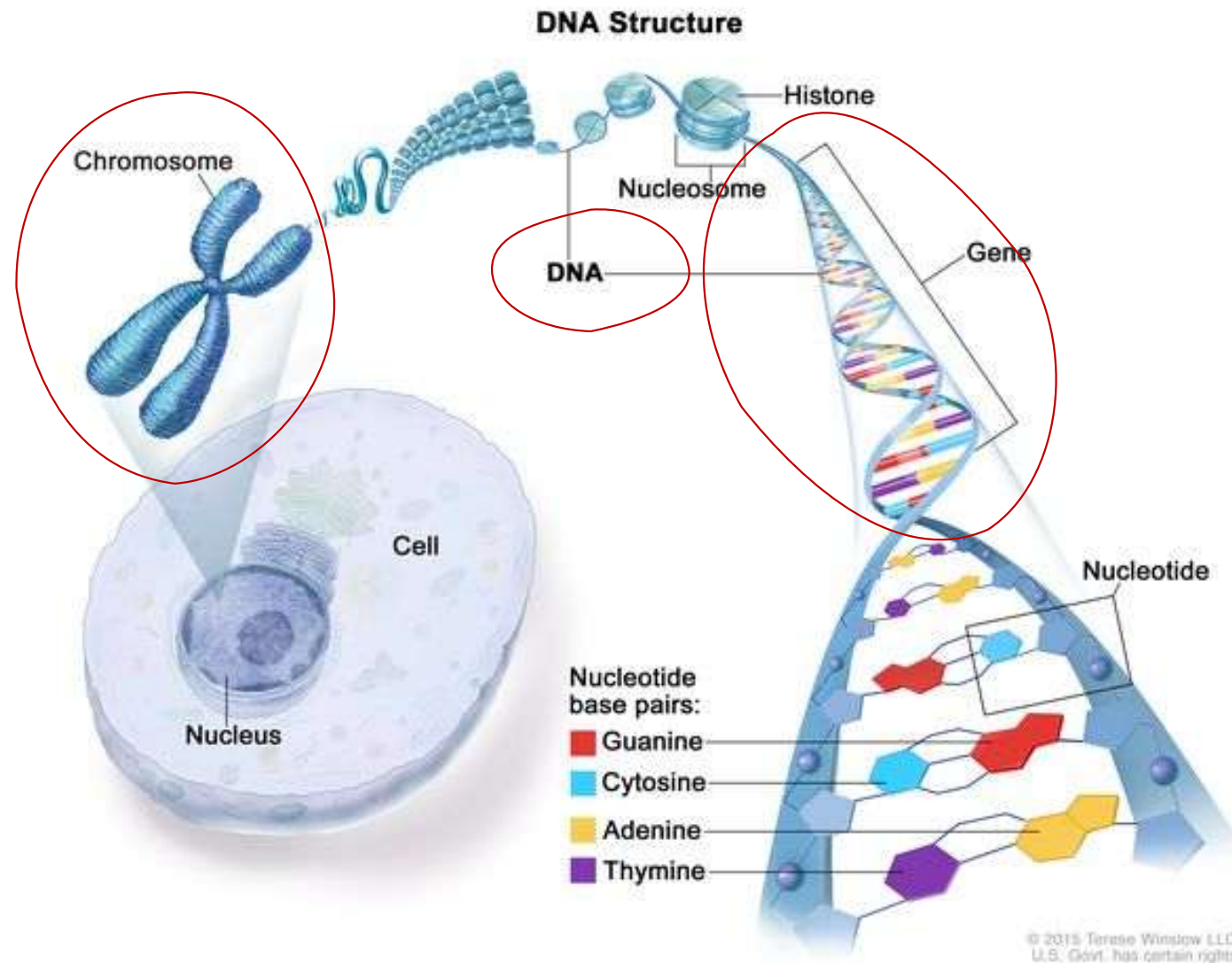
DNA, genes and chromosomes

DNA: Double helix containing the bases: Adenine, Thymine, Cytosine, Guanine

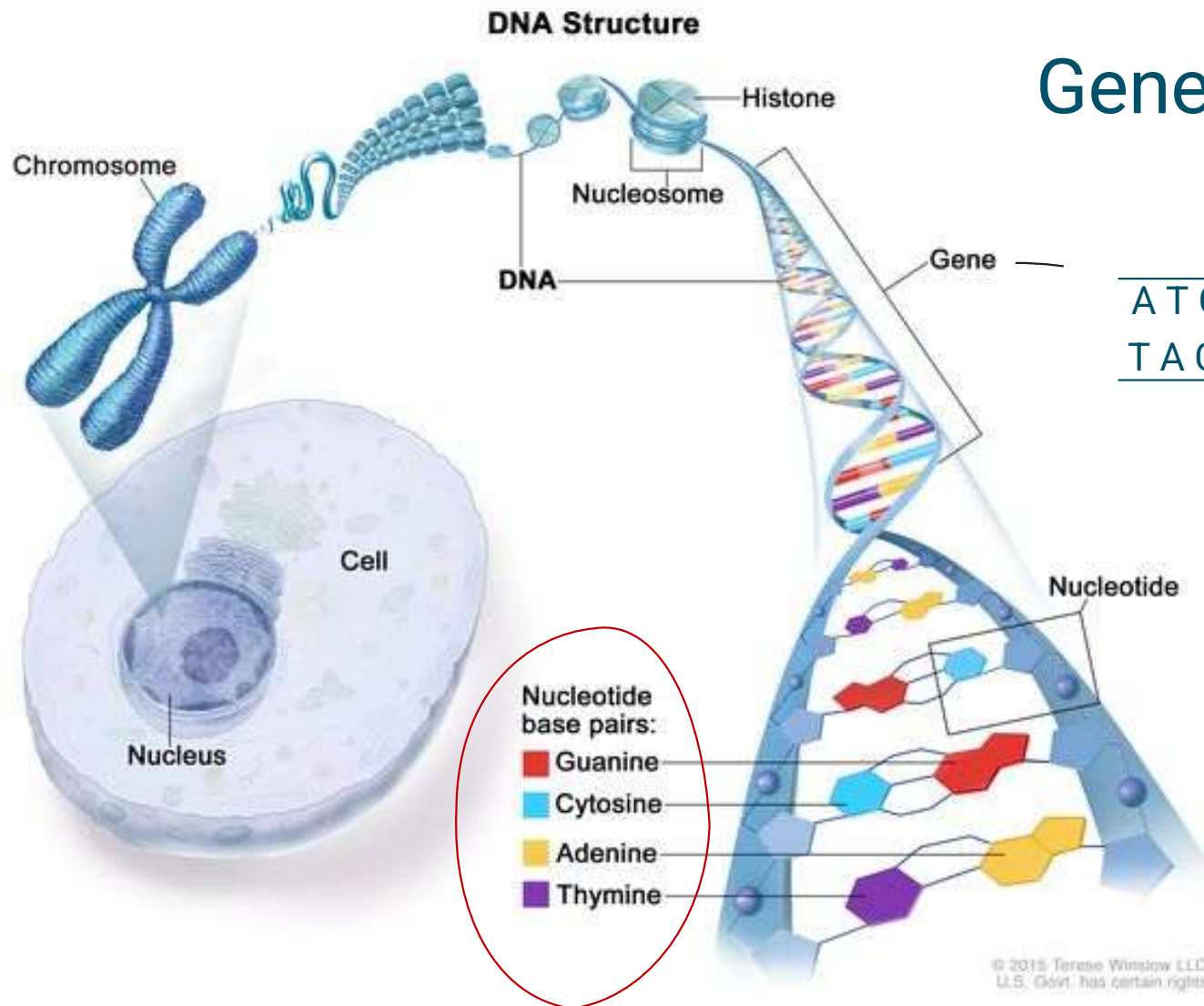
Chromosomes: Tightly coiled DNA

Genes: Small sections of DNA, coding proteins

Note: Most of the chromosomes are composed of DNA sequences that either lack a known function or whose function is not yet understood.



Genes and genetic data



ATGCTTACCGTAGTTTACTAA
TACAATGGCATCAAATGATCC

↑
Genetic data

Note: The term *genome* or *genomic data* refers to the complete set of genetic information in an organism.

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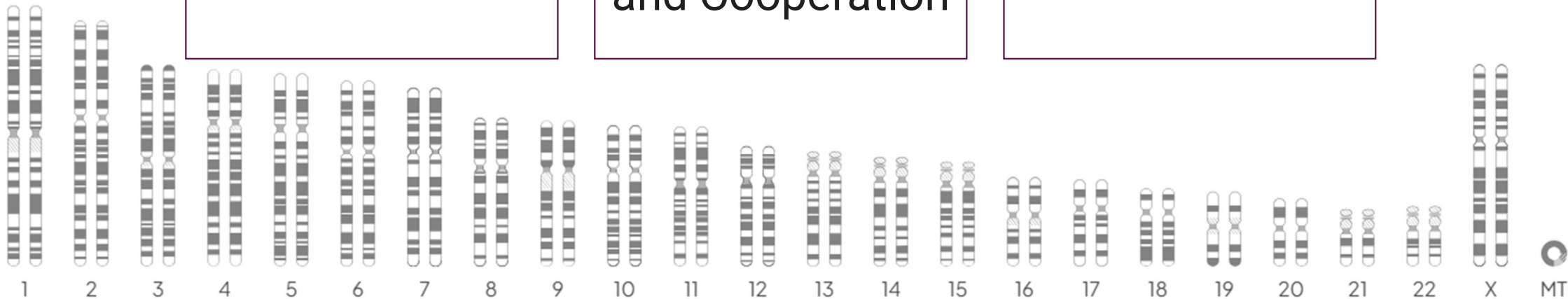
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Conditions

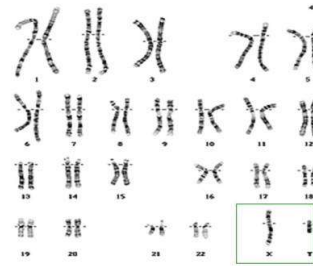
4. Medical
Genetics

5. IT,
Infrastructure
and Cooperation

6. Ethical
Considerations



1. DNA is extracted from blood samples or other biological material



2. The DNA is sequenced using sequencing machines



3. Data from the DNA are presented in different formats

RCV ID	Chr ↕	Position ↕	Variant ID	Gene ↕	Impact	Risk Frequency ↕	Ref	Alt	Your Data 📄	Risk Version	Your Status ⬆️	Condition ⬆️	Classification ⬆️	Confidence ⬆️	References	Links
RCV001275503	1	119422246	rs80358217	HSD3B2	p.Arg249Ter	0.0000948	C	T	CC	T	Harmless (NI)	3 Beta-Hydroxysteroid Dehydrogenase Deficiency ⓘ	Pathogenic	High (R)	ClinVar	Ensembl Viewer Geograp See all ▼
RCV003459912	1	119422184–119422211	rs2526392619	HSD3B2	p.Trp230_Ala238del		I	D	II	D	Harmless (NI)	3 Beta-Hydroxysteroid Dehydrogenase Deficiency ⓘ	Pathogenic	Medium (S)	ClinVar	Ensembl Viewer Geograp See all ▼
RCV000012967	1	119422013	rs80358216	HSD3B2	p.Trp171Ter	<0.00001	G	A	GG	A	Harmless (NI)	3 Beta-Hydroxysteroid Dehydrogenase Deficiency ⓘ	Pathogenic	Medium (S)	ClinVar	Ensembl Viewer Geograp See all ▼

DNA sequencing



Purpose

- To figure out the exact order of bases adenine, thymine, cytosine, and guanine (A, T, C, G) in a DNA molecule.

How is it done

- Example video: [Overview of Illumina Sequencing by Synthesis Workflow \(youtube.com\)](https://www.youtube.com/watch?v=7F000000000)

Formats of resulting files

- FASTQ, BAM/SAM, VCF

Rapid Development in Sequencing Technology

- Faster sequencing machines
- Lower costs
- Higher quality, and more detailed data
- Studying more parts of the DNA
 - Example: the whole genome and how genes are regulated (epigenetics)

Source: <https://www.genomicseducation.hee.nhs.uk/education/core-concepts/what-is-epigenetics>.
Screenshot from <https://allhealthtech.com/fast-cheap-genome-sequencing-novaseq-x-series>

The screenshot shows a webpage from Allhealthtech, dated November 10, 2022. The article title is "Genome Sequencing: More insights faster and cheaper with NovaSeq X series". The text states: "Illumina unveiled the NovaSeq X series, the fastest and most cost-efficient genome sequencing machine. Know more here." Below the text is a large image of the NovaSeq X Series machine, a tall, silver, industrial-looking device. To the right of the machine is a graphic of a DNA double helix. Text overlaid on the image reads: "illumina NovaSeq X Series The fastest and cheapest genome sequencing machine". At the bottom right of the image, a red text box says: "NovaSeq X series can crack human genomes twice as fast as its current models, bringing down the costs from \$600 per person to a mere \$200 per person!". At the bottom of the screenshot, there is a quote: "Genetic disorders can't be cured no matter what you do!" - A statement you might be tired of hearing, which may soon be untrue. Yes, you read that right!

Core file formats

FASTQ

- Text-based format
- Stores raw reads with quality scores

```
@SEQ_ID_1
ACGTGACCTGA
+
IIIIIIIIII
```

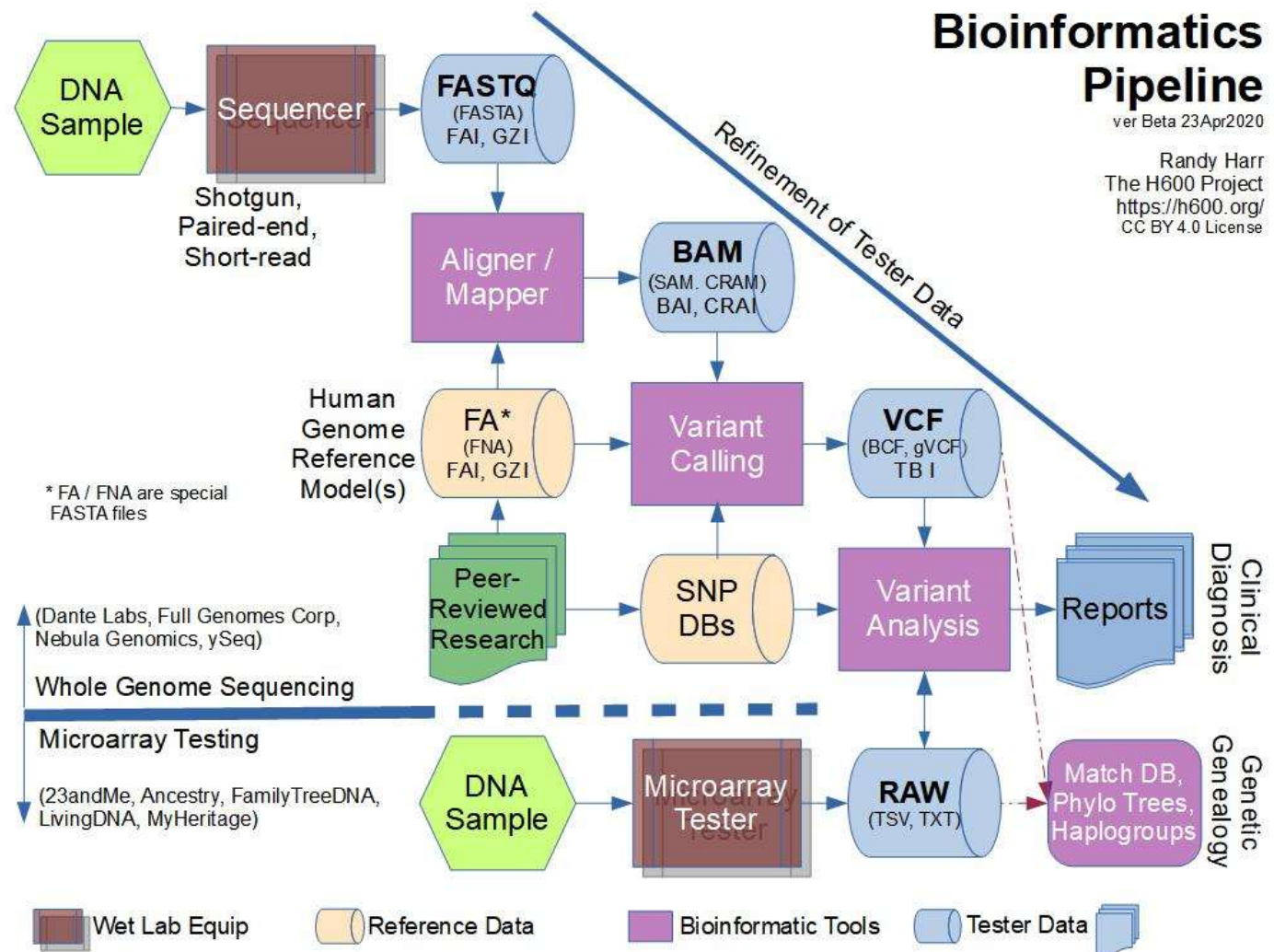
BAM

- Stores reads aligned to a reference genome
- Compressed, binary format, efficient for large datasets
- Not human-readable directly (need tools like samtools to view it)

VCF

- A text format that stores genetic variants (differences from a reference genome), such as SNPs or insertions.

```
#CHROM POS ID REF ALT QUAL FILTER INFO
chr1 105 . G A 99 PASS DP=30
```



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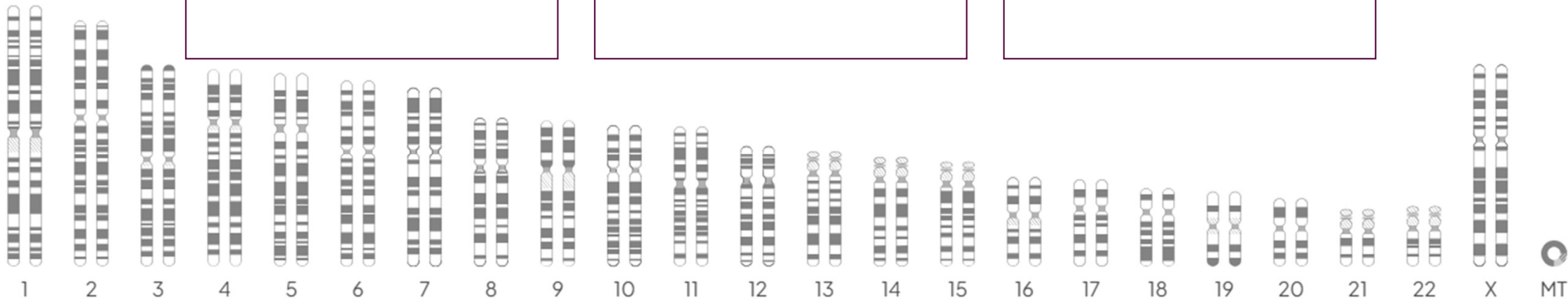
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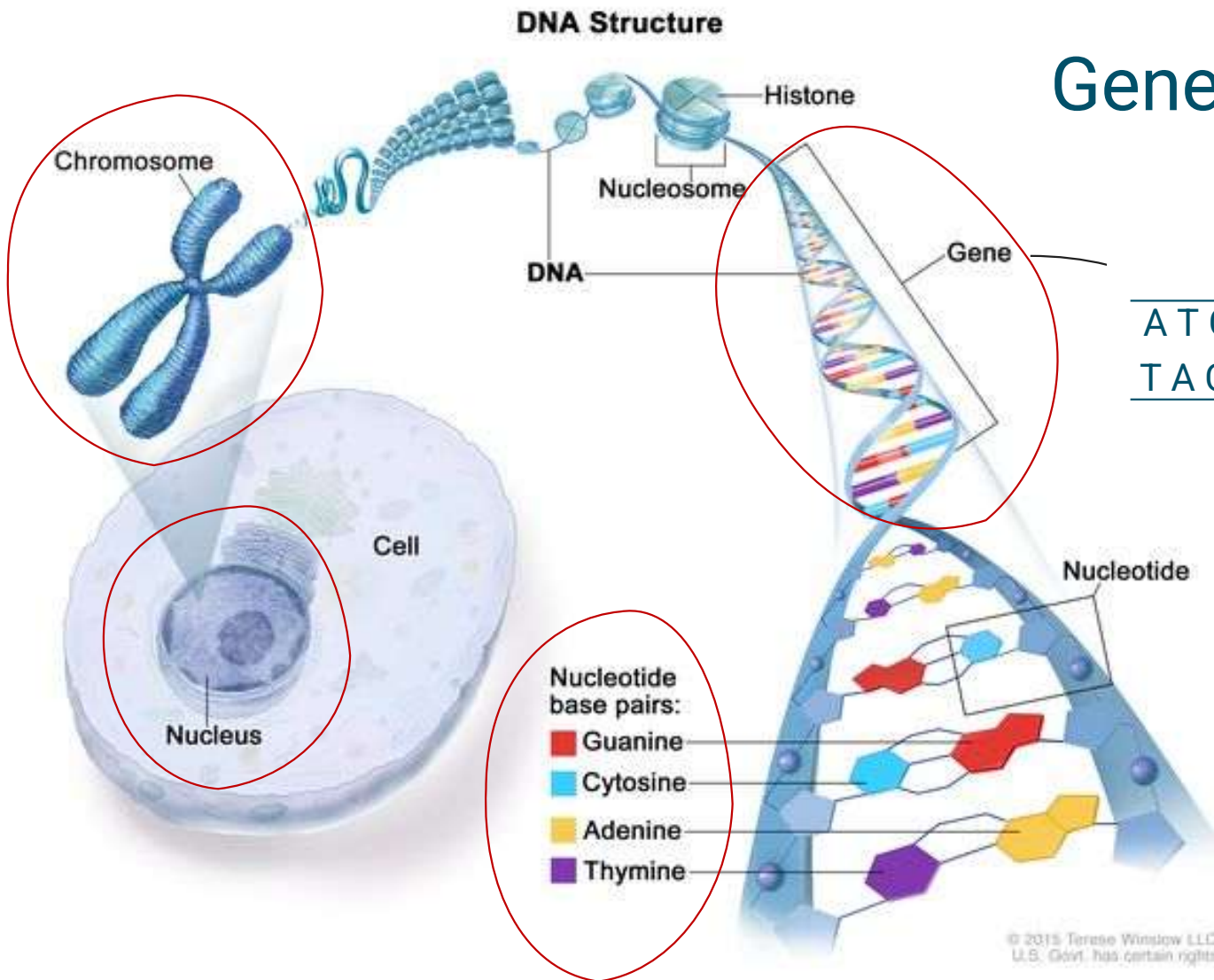
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Genes and genetic data - summary



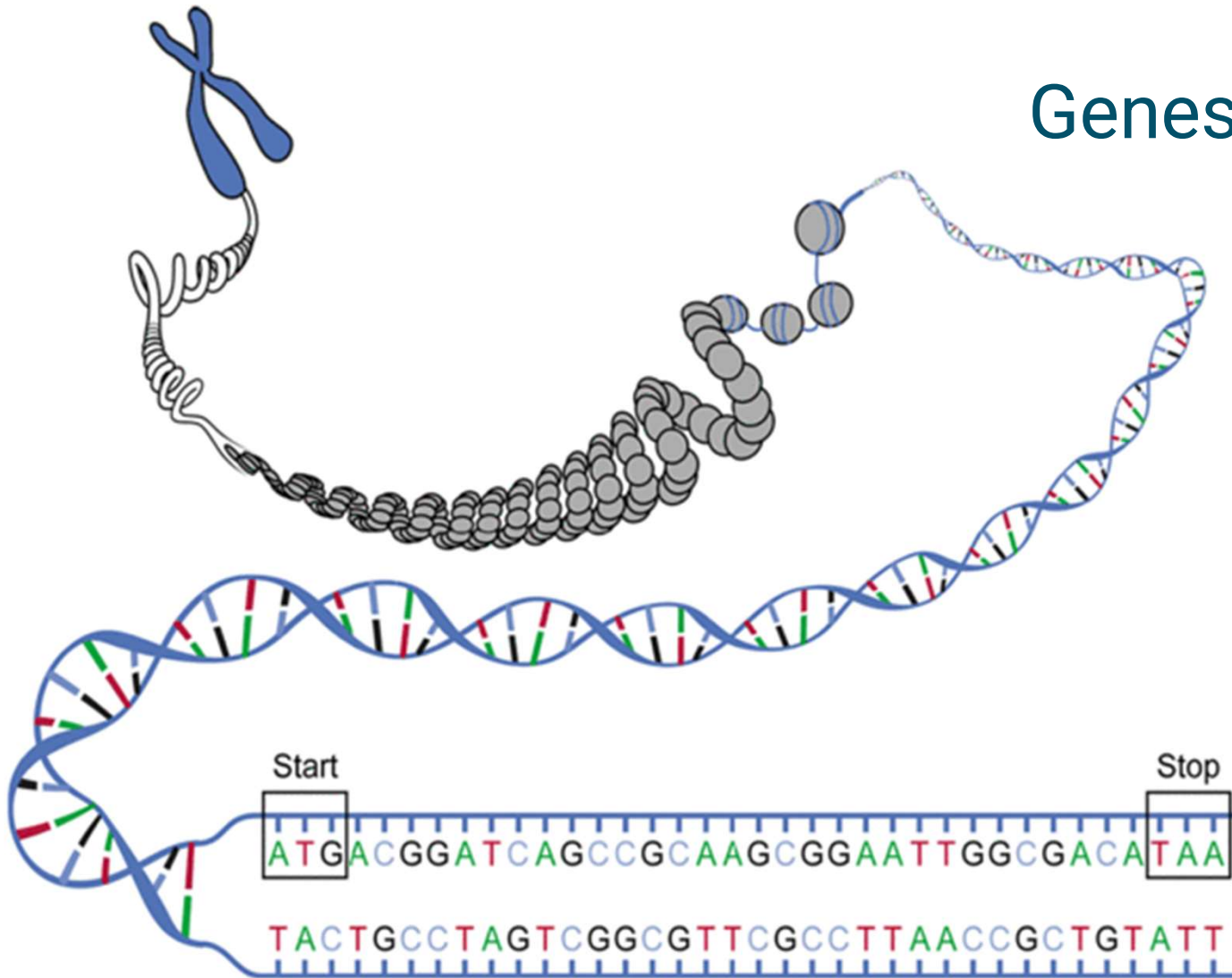
ATGCTTACCGTAGTTTACTAA
TACAATGGCATCAAATGATCC

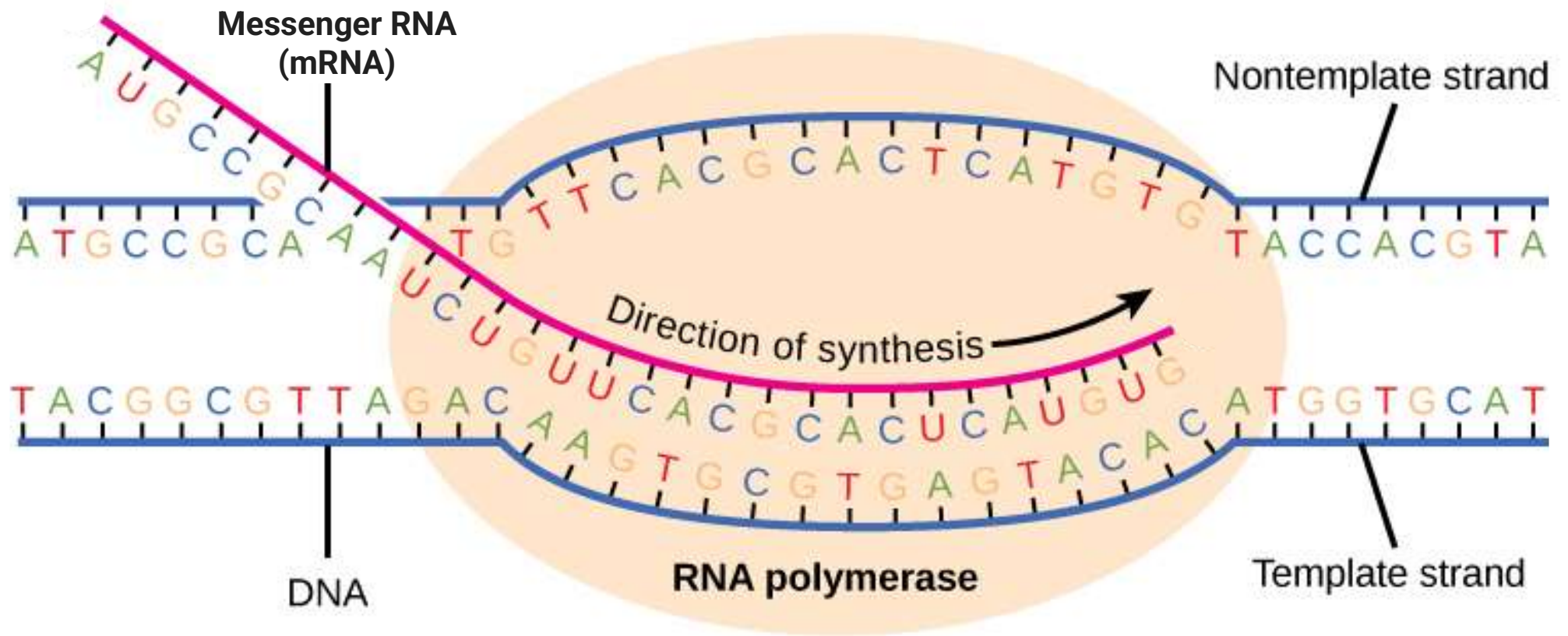
↑
Genetic data

Genes and genetic data

Length of a gene:
Between a few hundred
base pairs and over 2,4
million base pairs.

Note: The reading of a
gene starts at the “start
codon”, and ends at the
“stop codon”

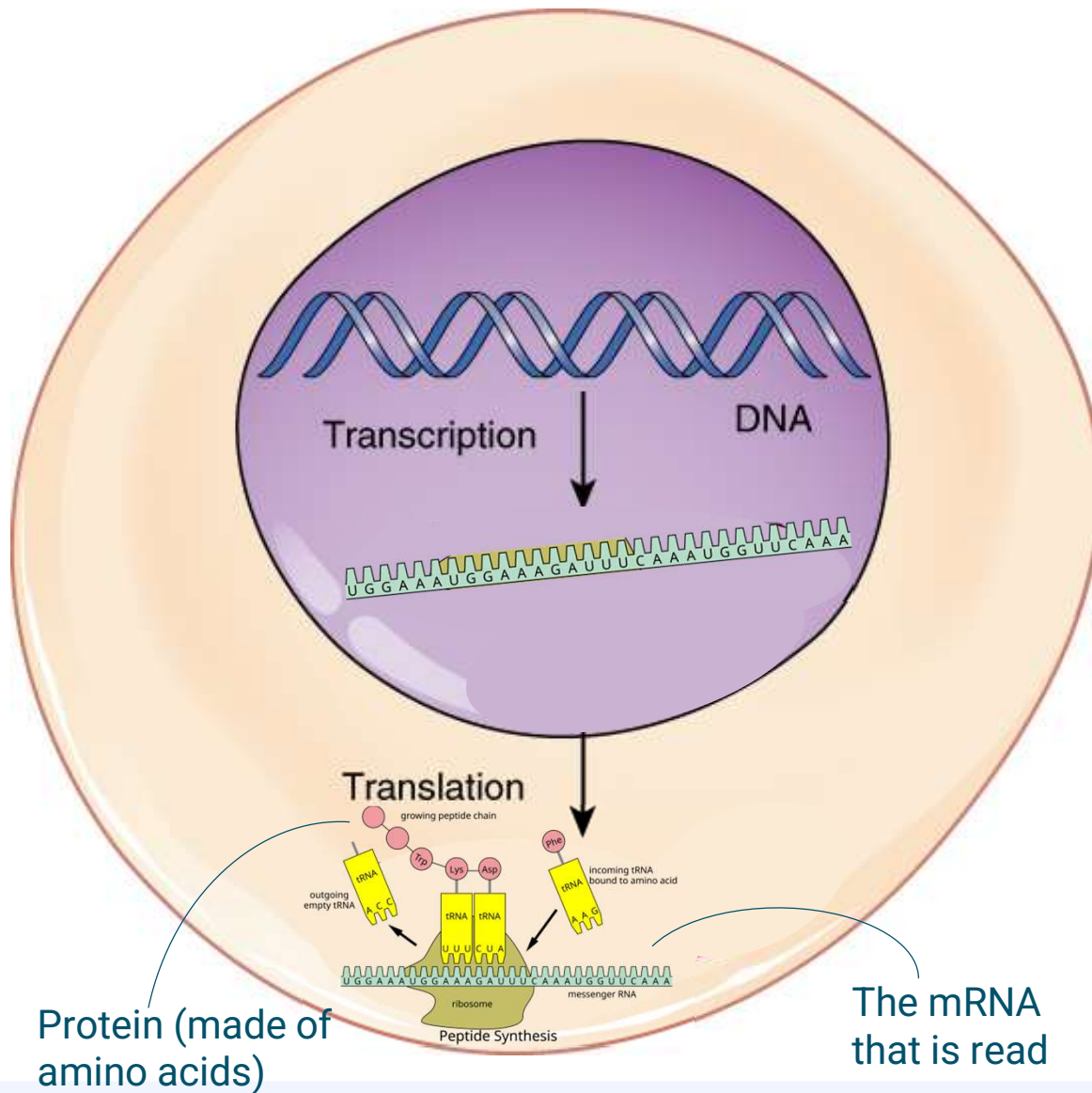


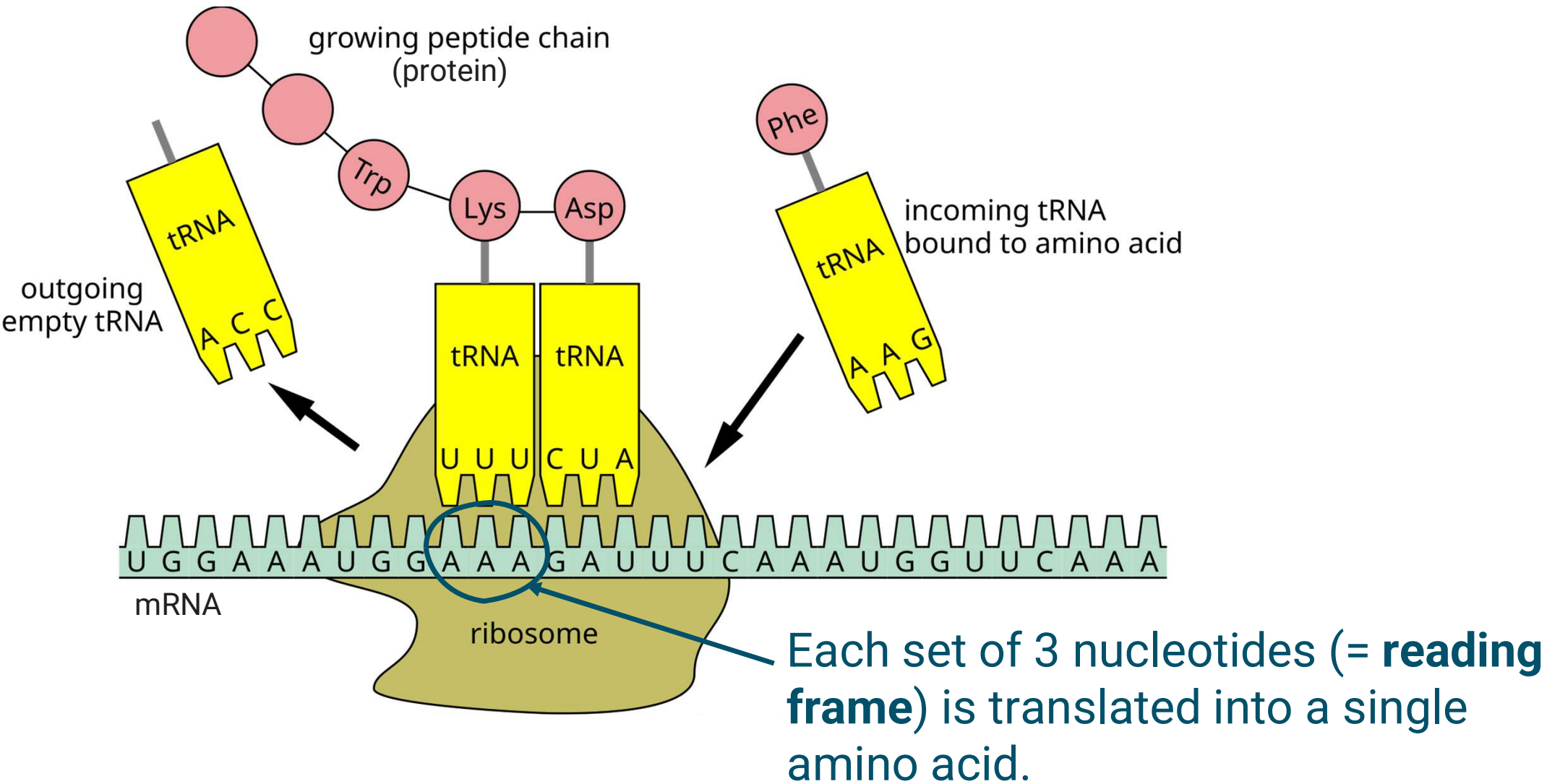


Gene Expression

Gene expression = turning genetic information into something the cell can use:

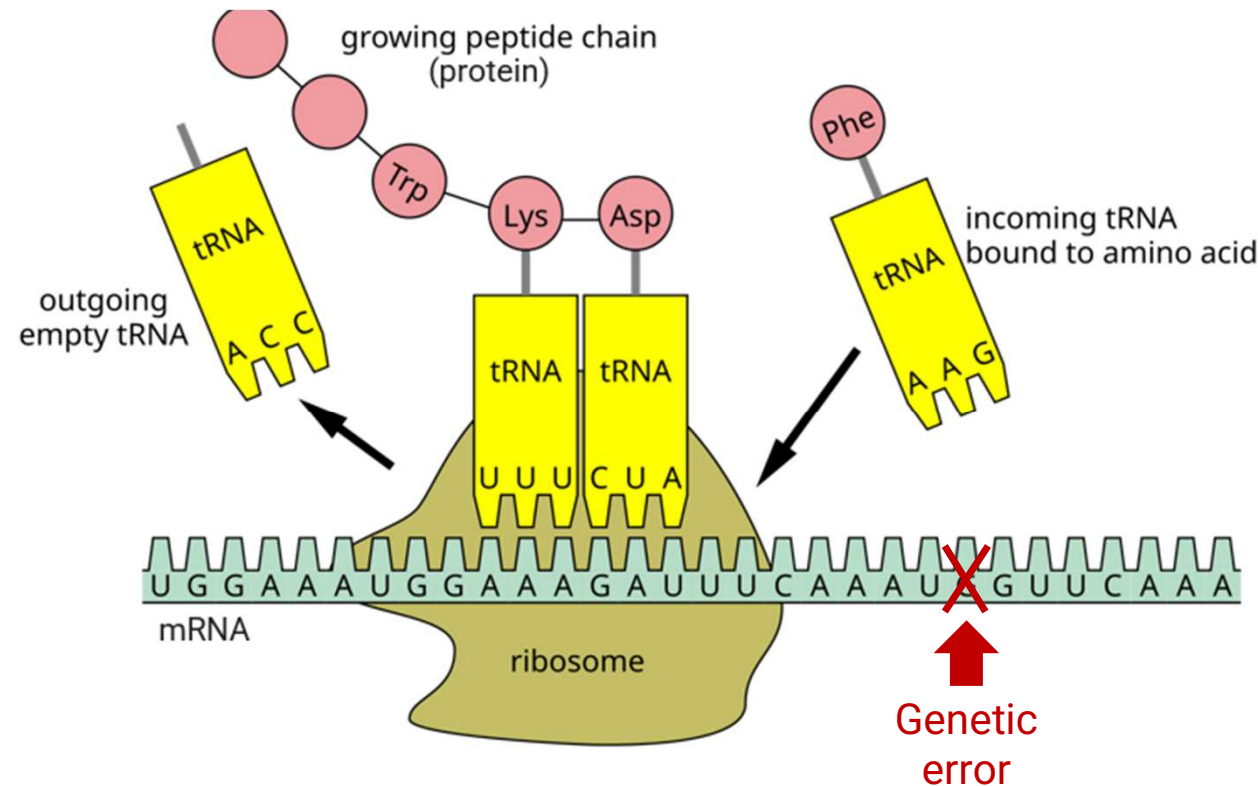
1. The cell copies a gene from DNA into mRNA
2. mRNA leaves the nucleus
3. mRNA is used as a template to build a protein

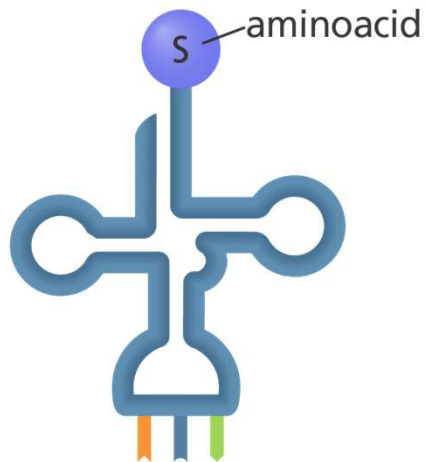




Genetic errors (mutations)

- can occur when cells copy their DNA, or due to external factors such as radiation, chemicals, or some viruses
- can be inherited, or they can arise during life
- are often repaired by the cell's DNA repair systems
- occur frequently in all individuals, but only a small number lead to disease or increased disease risk
- can be identified through genetic testing, which may reveal mutations linked to certain diseases





**transfer RNA
(tRNA)**

The nucleotides
found in RNA:

U = Uracil

C = Cytosine

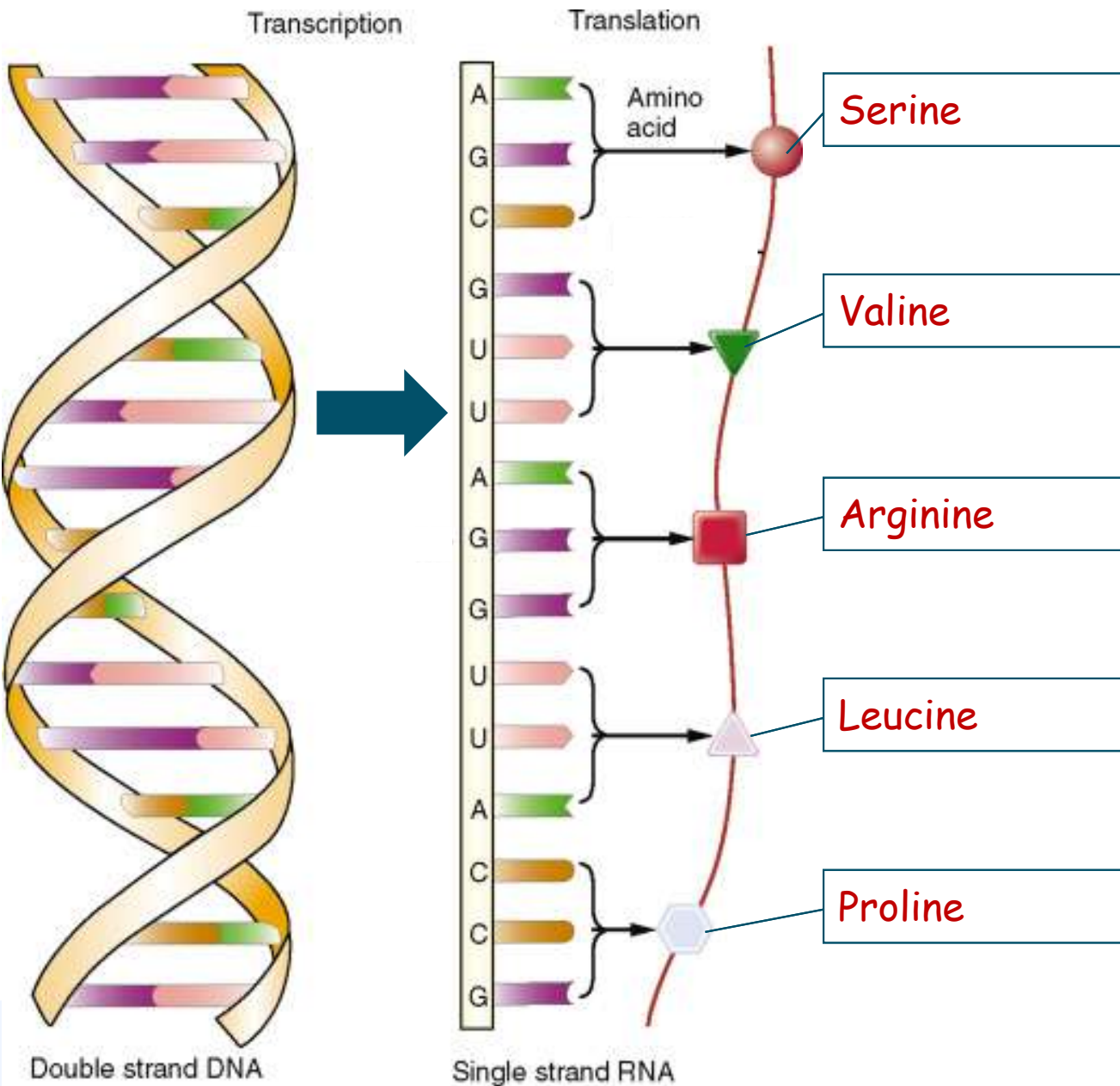
A = Adenine

G = Guanine

RNA uses uracil (U)
instead of thymine
(T), which is used in
DNA.



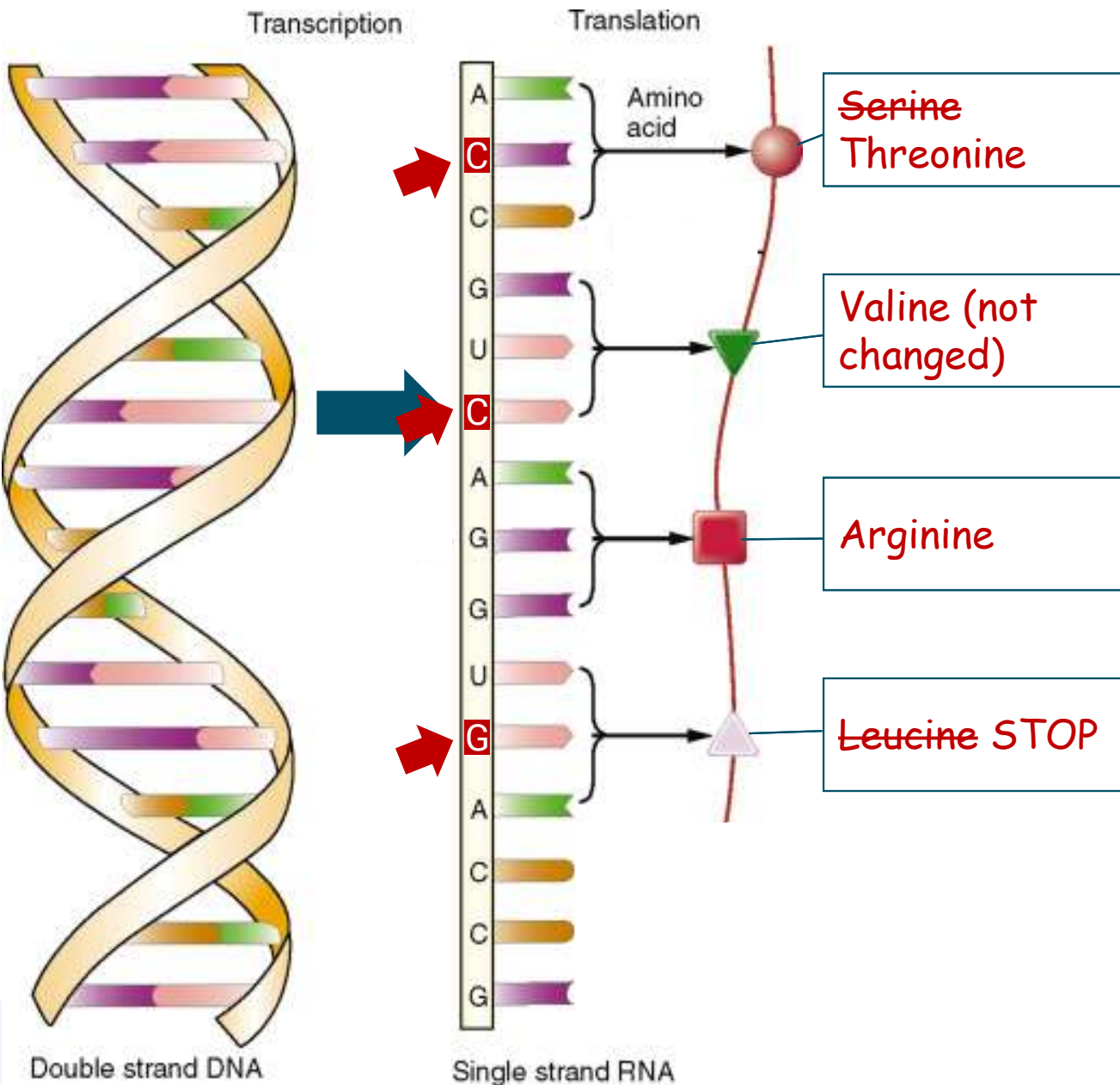
First Base in the Codon	Second Base of the Codon				Third Base in the Codon
	U (T)	C	A	G	
U (T)	UUU Phenylalanine	UCU Serine	UAU Tyrosine	UGU Cysteine	U (T) C A G
	UUC Phenylalanine	UCC Serine	UAC Tyrosine	UGC Cysteine	
	UUA Leucine	UCA Serine	UAA STOP	UGA STOP	
	UUG Leucine	UCG Serine	UAG STOP	UGG Tryptophan	
C	CUU Leucine	CCU Proline	CAU Histidine	CGU Arginine	U (T) C A G
	CUC Leucine	CCC Proline	CAC Histidine	CGC Arginine	
	CUA Leucine	CCA Proline	CAA Glutamine	CGA Arginine	
	CUG Leucine	CCG Proline	CAG Glutamine	CGG Arginine	
A	AUU Isoleucine	ACU Threonine	AAU Asparagine	AGU Serine	U (T) C A G
	AUC Isoleucine	ACC Threonine	AAC Asparagine	AGC Serine	
	AUA Isoleucine	ACA Threonine	AAA Lysine	AGA Arginine	
	AUG Methionine	ACG Threonine	AAG Lysine	AGG Arginine	
G	GUU Valine	GCU Alanine	GAU Aspartic Acid	GGU Glycine	U (T) C A G
	GUC Valine	GCC Alanine	GAC Aspartic Acid	GGC Glycine	
	GUA Valine	GCA Alanine	GAA Glutamic Acid	GGA Glycine	
	GUG Valine	GCG Alanine	GAG Glutamic Acid	GGG Glycine	



Excercise 1

- What amino acids are produced by the following RNA strand?
- Use the codon table

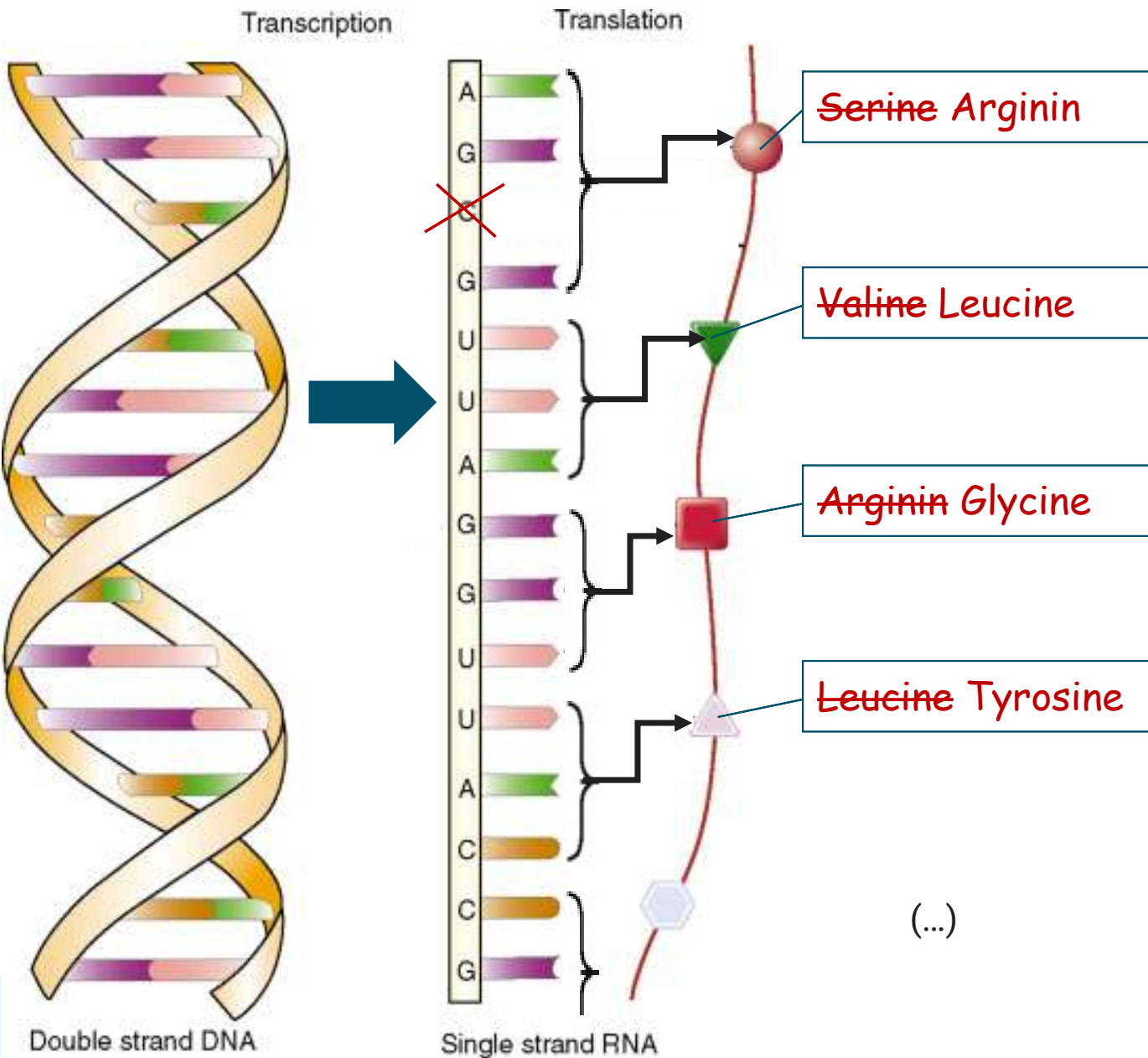
(answer in **red**)



Excercise 2

- What amino acids are produced by the following RNA strand, containing errors?
- Use the codon table

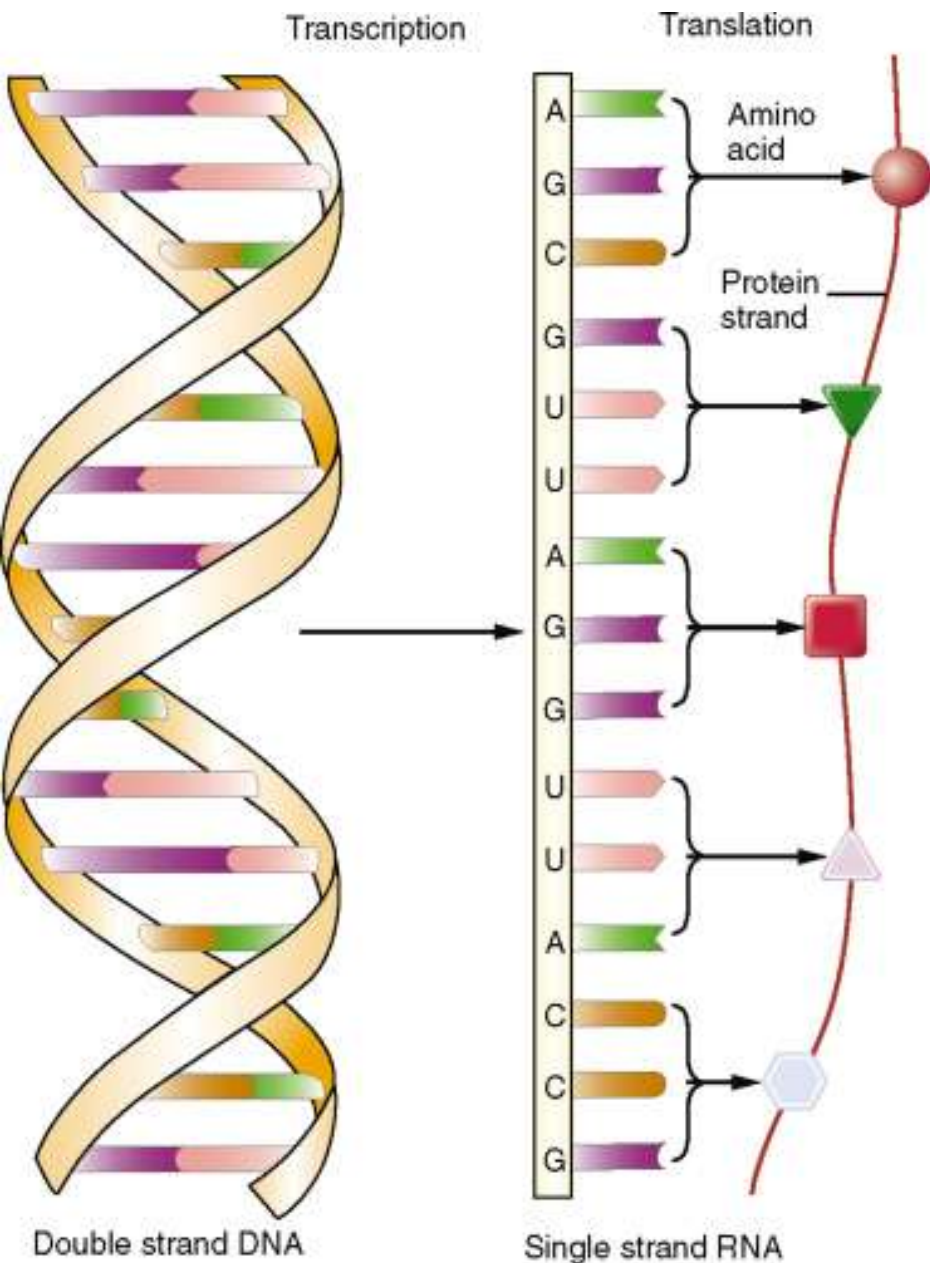
(answer in **red**)



Excercise 3

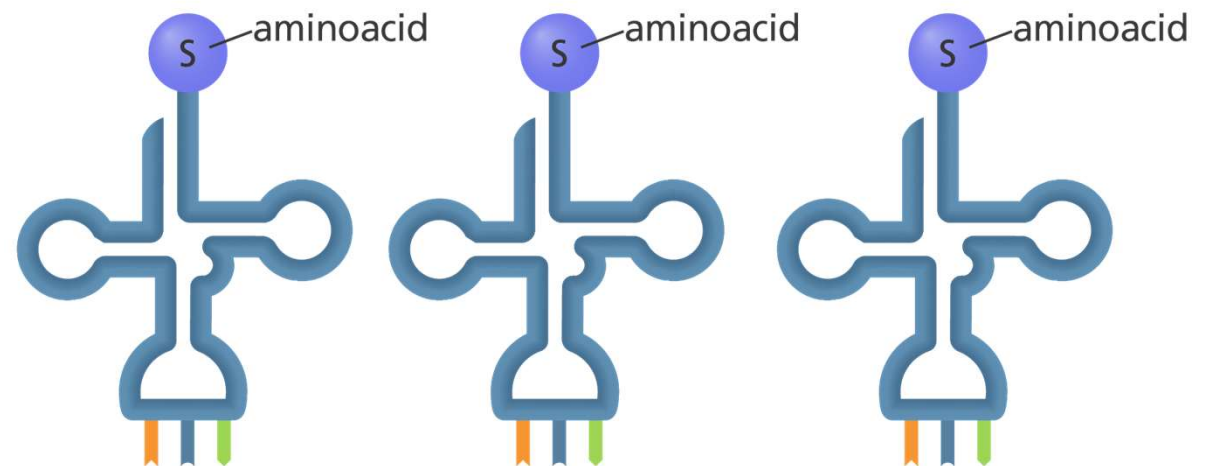
- What amino acids are produced by the following RNA strand, when a nucleotide is missing?
- Use the codon table

(answer in **red**)



Frameshift mutation

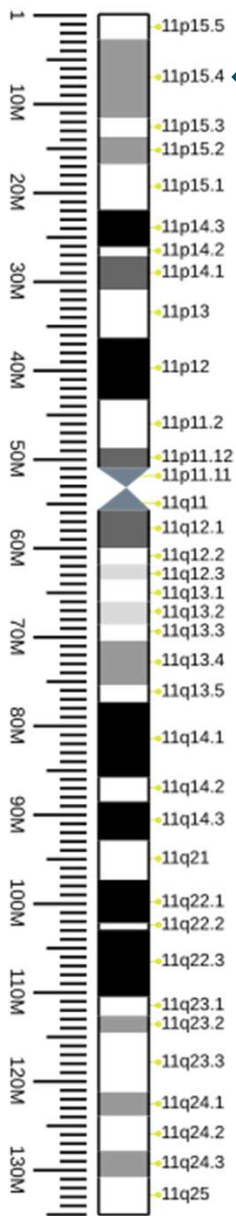
Frameshift mutation is when an insertion or deletion changes the entire reading frame of a gene.



What are the consequences of the change?

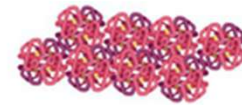
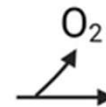
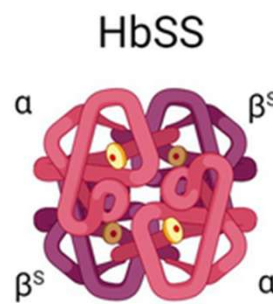
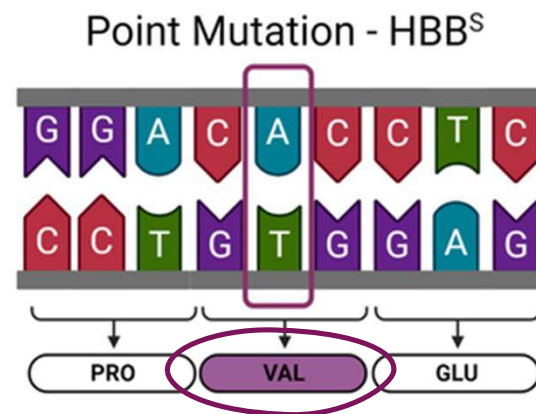
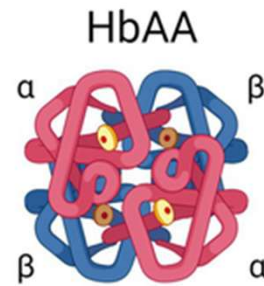
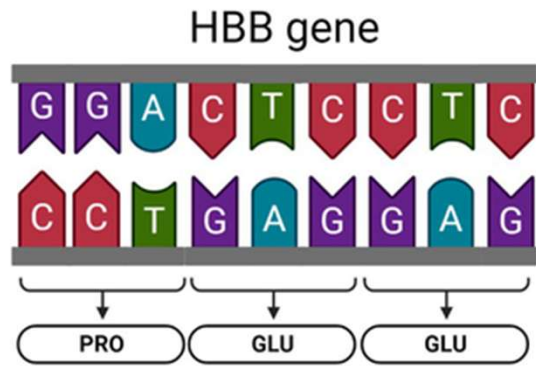
From no effect to big consequences

- Diseases
- Developmental disorders
- Increased disease risk
- Changed response on medication (called pharmacogenetics)

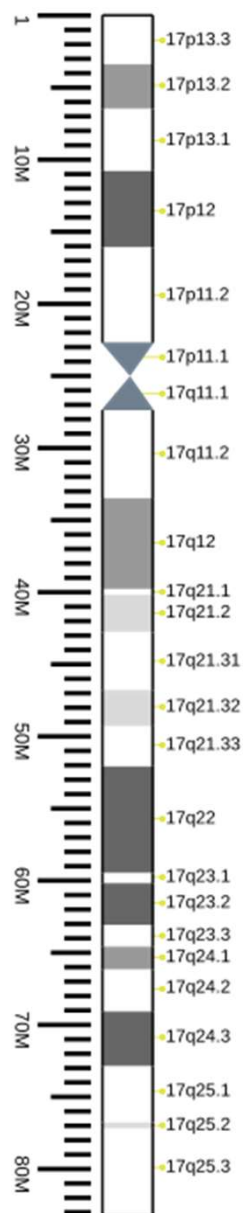


HBB

Point mutation (single nucleotide change)



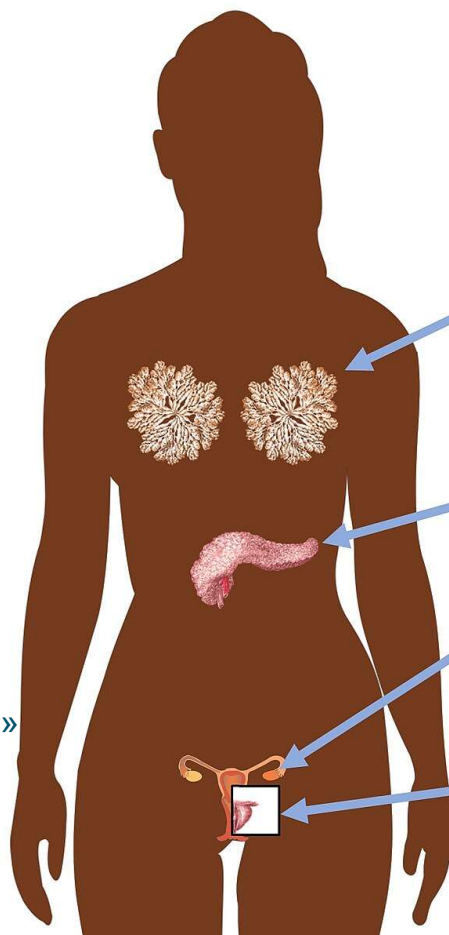
Sickle cell anemia



BRCA1

Duplication

Called «BRCA1 c.5266dup»
(p.Gln1756fs)



Breast cancer:

BRCA1

50% to 65%

Males: 1.2%

BRCA2

40% to 55%

Males: Up to 9%

Pancreas cancer:

1-3%

2-7%

Ovarian cancer:

40% to 65%

15% to 25%

Prostate cancer:

9%

15%

Petrucelli N, Daly MB, Pal T (September 1998). "[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)". In Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJ, Gripp KW, Amemiya A, editors (eds.). *GeneReviews® [Internet]*. Seattle (WA): University of Washington, Seattle. [PMID 20301425](#).

Chromosomal mutations

Cri-du-chat

Partial deletion on chr 5



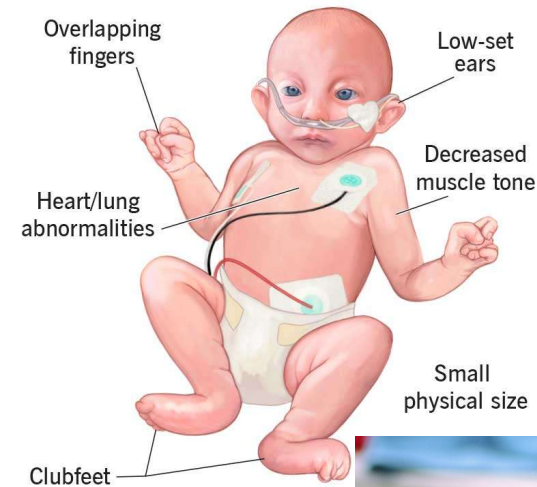
William syndrome

Partial deletion on chr 7



Edward's syndrome

3 copies of chr 18



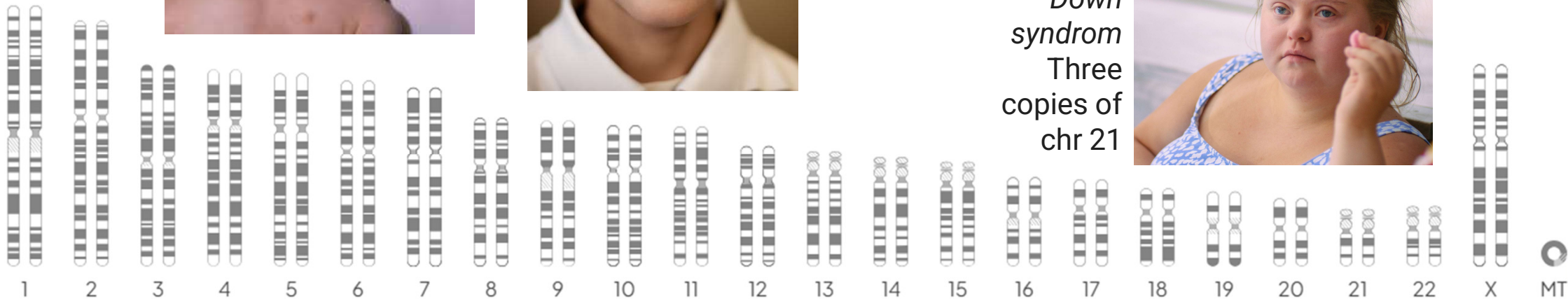
Turner syndrome

Only one copy of chr X



Down syndrom

Three copies of chr 21



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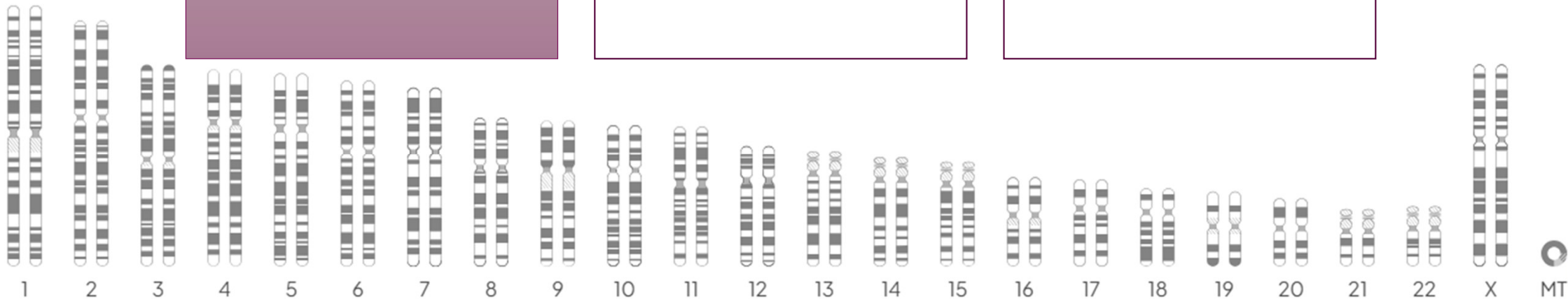
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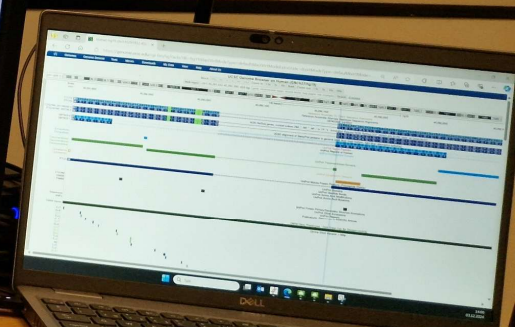
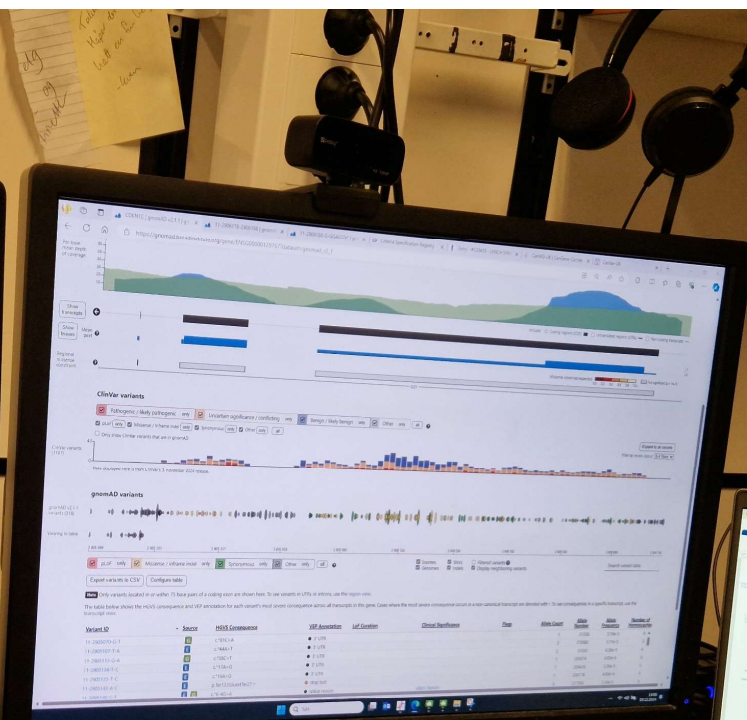
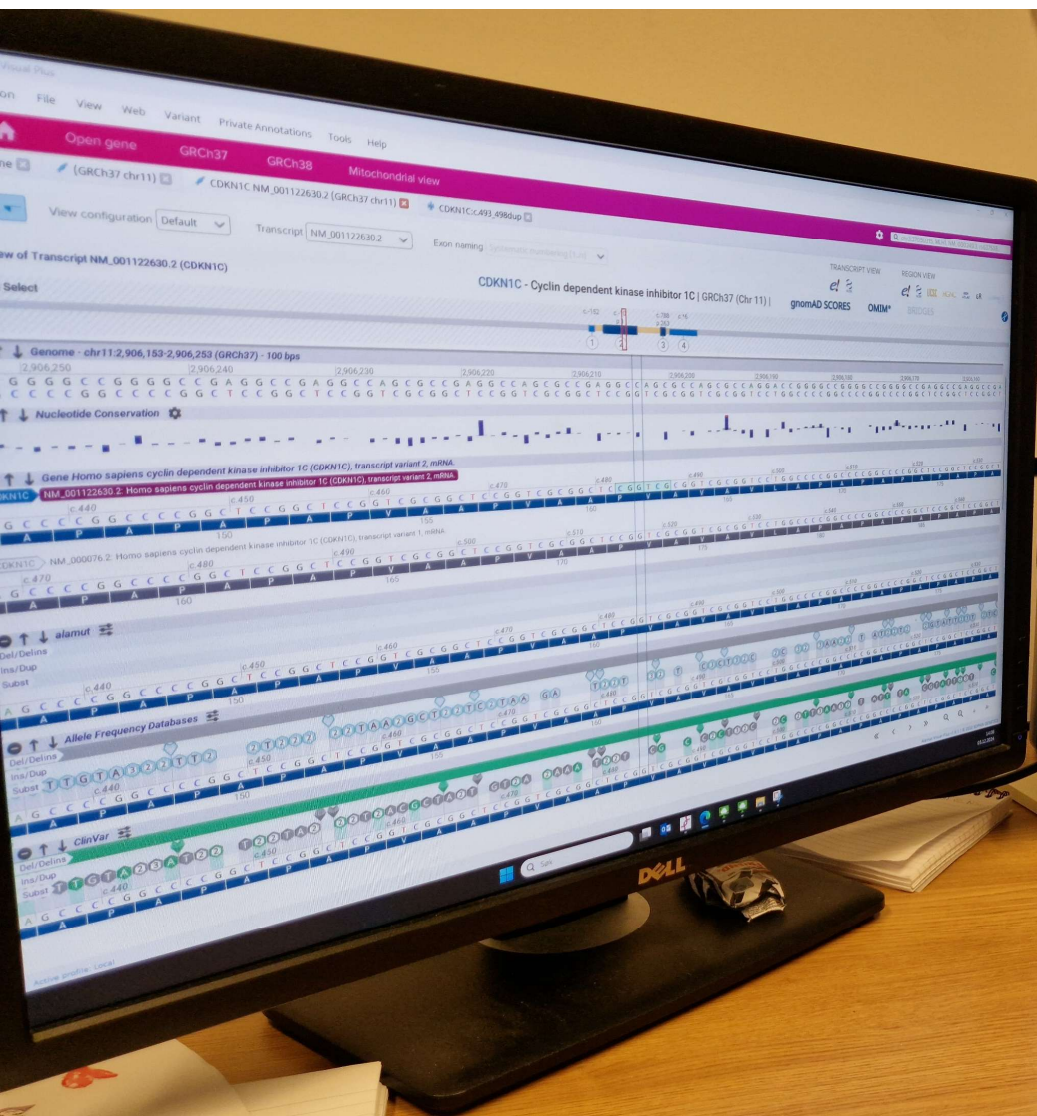
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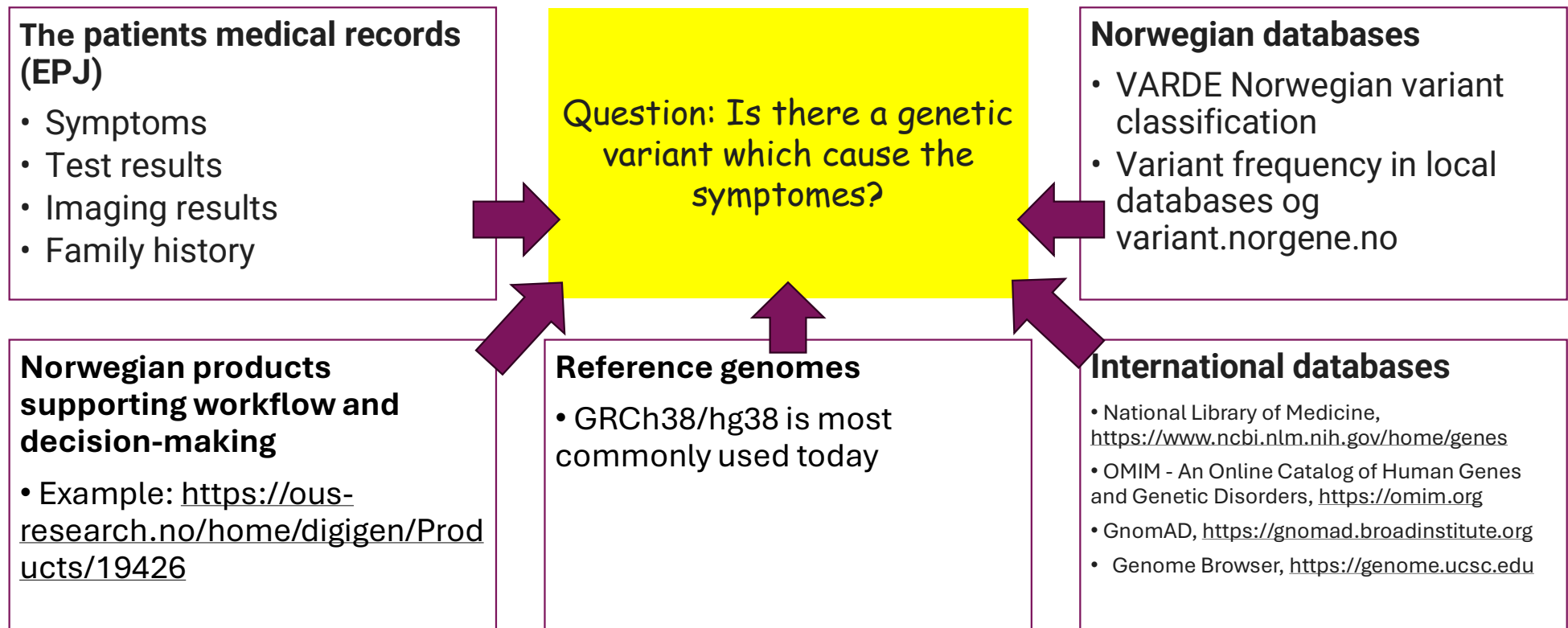
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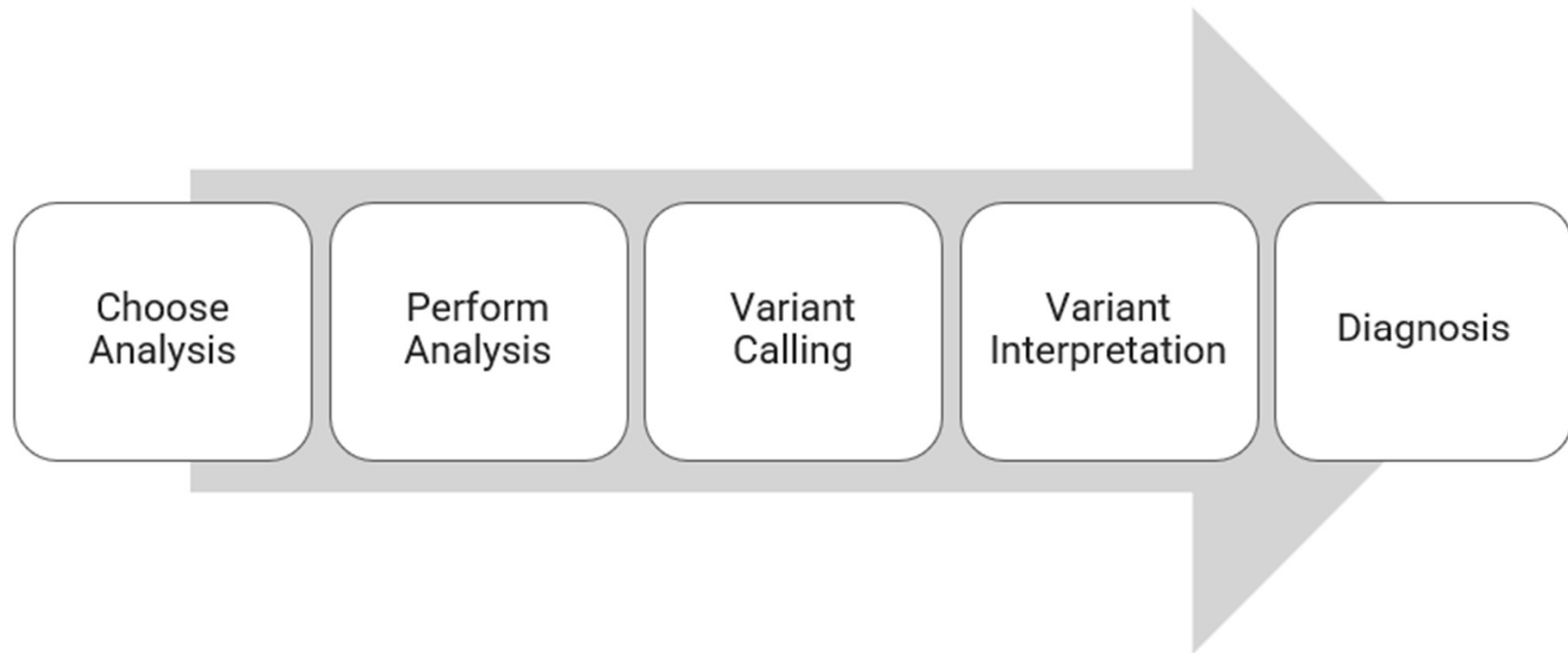




Tools and Databases Supporting Genetic Testing



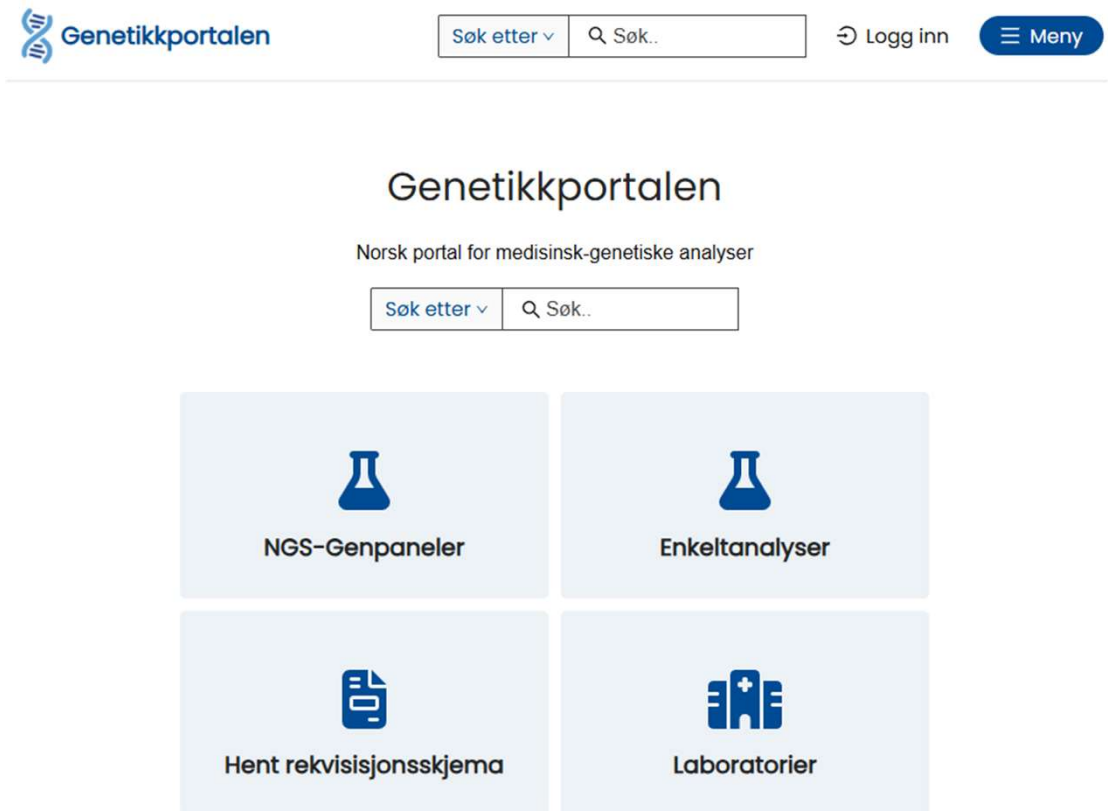
Workflow at the Lab of Medical Genetics



Workflow at the Lab of Medical Genetics:

1) Choose right analysis

- Choose parts of the DNA to analyse («panels»)
- Many of the panels used are described at [Genetikkportalen](https://genetikkportalen.no)
- Examples of additional data which might be helpful for the hospital:
 - The patient's symptoms, age, sex
 - Suspected condition
 - Results of relevant blood tests, radiological images
 - Related diseases in family



Workflow at the Lab of Medical Genetics:

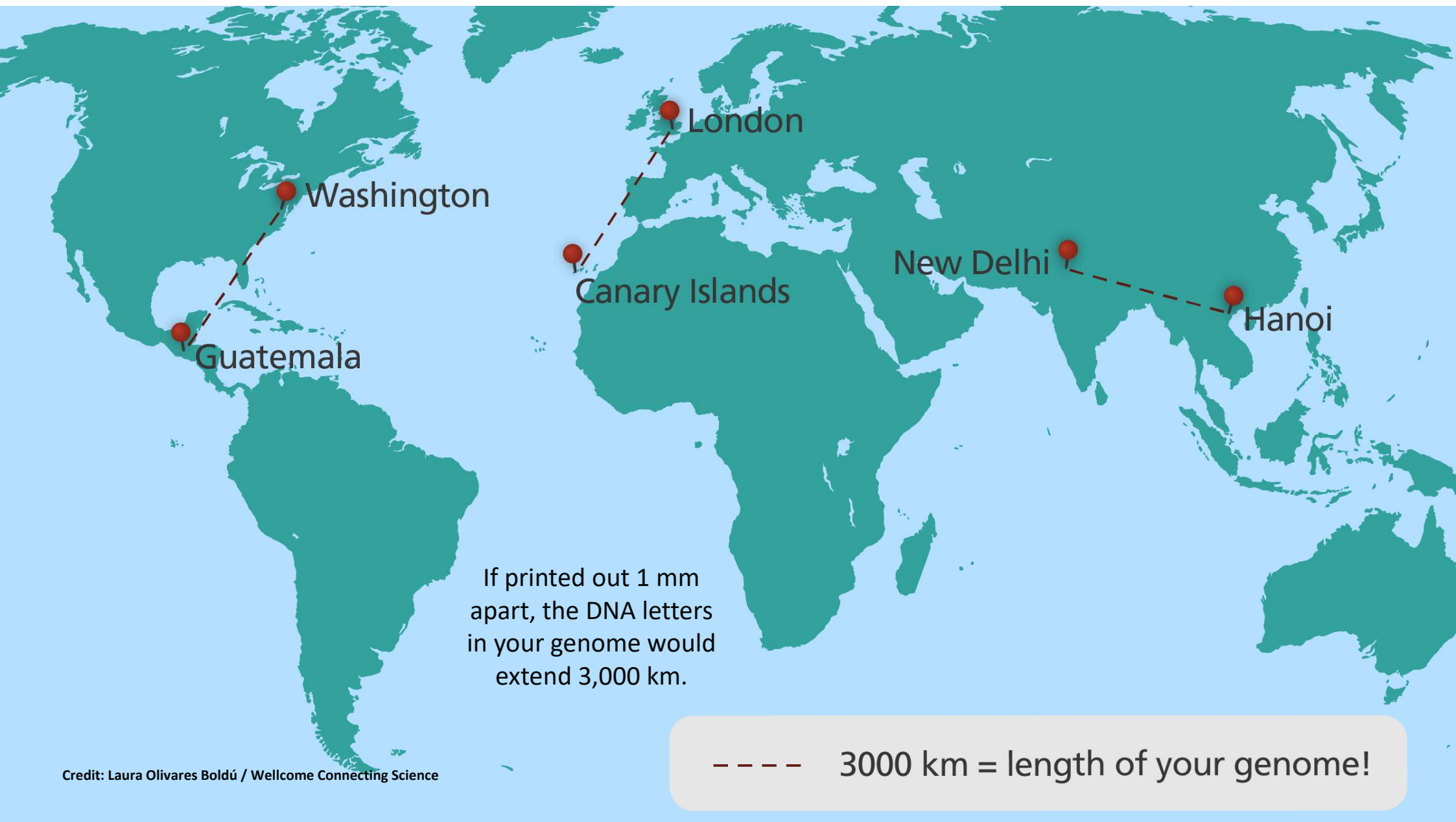
2) Perform analysis



Example: Sequencing-by-synthesis (Illumina)

The entire human genome consists of more than 3 billion base pairs (3×10^9) and contains around 20,000–25,000 genes.

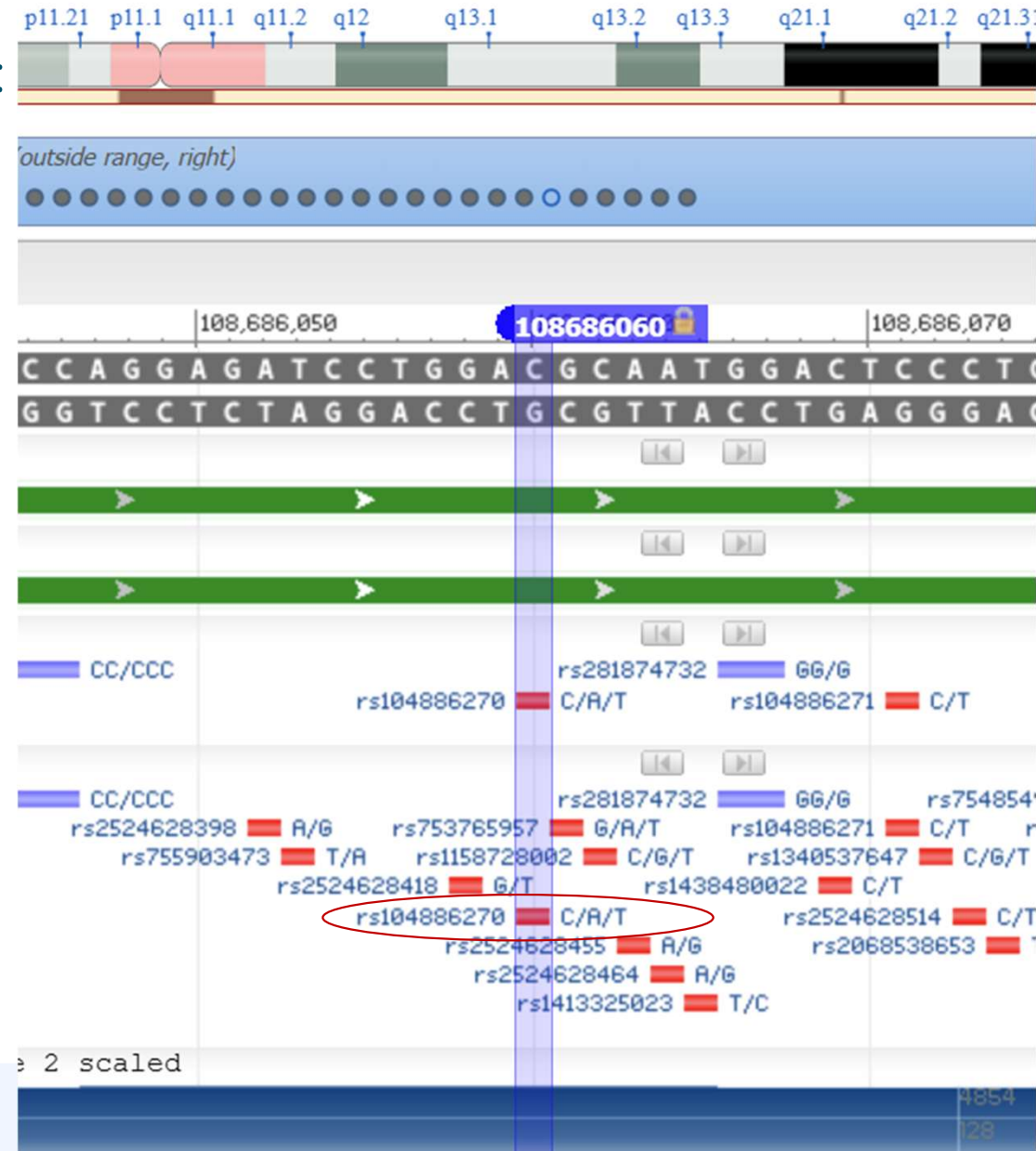
One genome is typically 100 gigabytes of raw data.

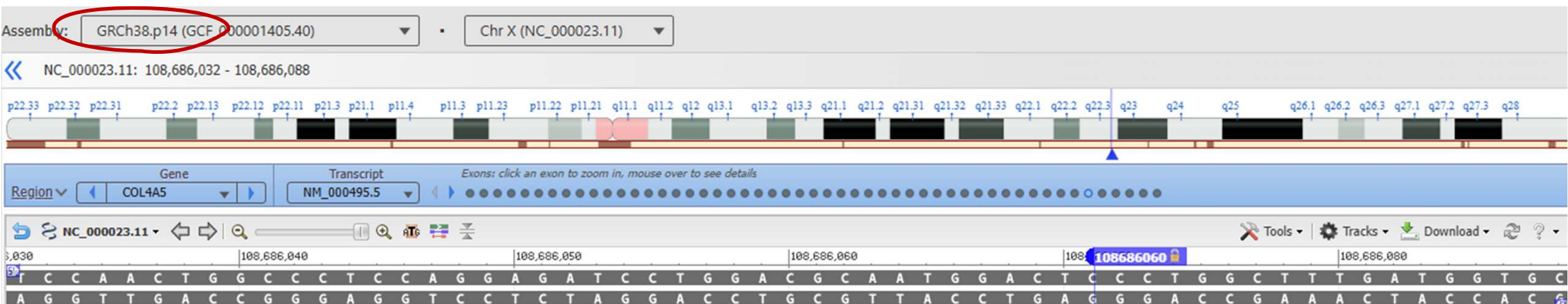


Workflow at the Lab of Medical Genetics:

3) Variant calling

- “Variant calling” is the process of identifying differences in DNA sequences between a sample and a reference genome.
- The sequenced DNA is aligned (matched) to a known reference genome
- During this comparison, genetic variants are found





A **reference genome** is a digital DNA sequence used as a standard to compare individual genomes.

- **Human genome 38 (hg38)** or GRCh38 is the current standard in most contexts
- GRCh37 (hg19) is still used in older tools/datasets
- T2T-CHM13 is a groundbreaking full genome, including previously missing parts, but is not yet fully adopted in clinical practice

Genome Data Viewer

Homo sapiens (human)

Search assembly
Q X:108686060

Assembly: GRCh38.p14 (GCF_000001405.40) Chr X (NC_000023.11)

NC_000023.11: 108,686,032 - 108,686,088

Gene: COL4A5 Transcript: NM_000495.5

Region: COL4A5

NC_000023.11: 108,686,040 108,686,050 108,686,060 108,686,070 108,686,080

NCBI RefSeq Annotation GCF_000001405.40-RS_2024_08

Genes, Ensembl release 113

Cited Variations, dbSNP b157 v2

Live RefSNPs, dbSNP b157 v2

RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation Release 110

Filtered (read count from min to max)

Loading... NC_000023.11: 109M..109M (57 nt)

Example of variant notation:
X-108686060-C-T

Resolves into the variant
RS104886271

Workflow at the Lab of Medical Genetics:

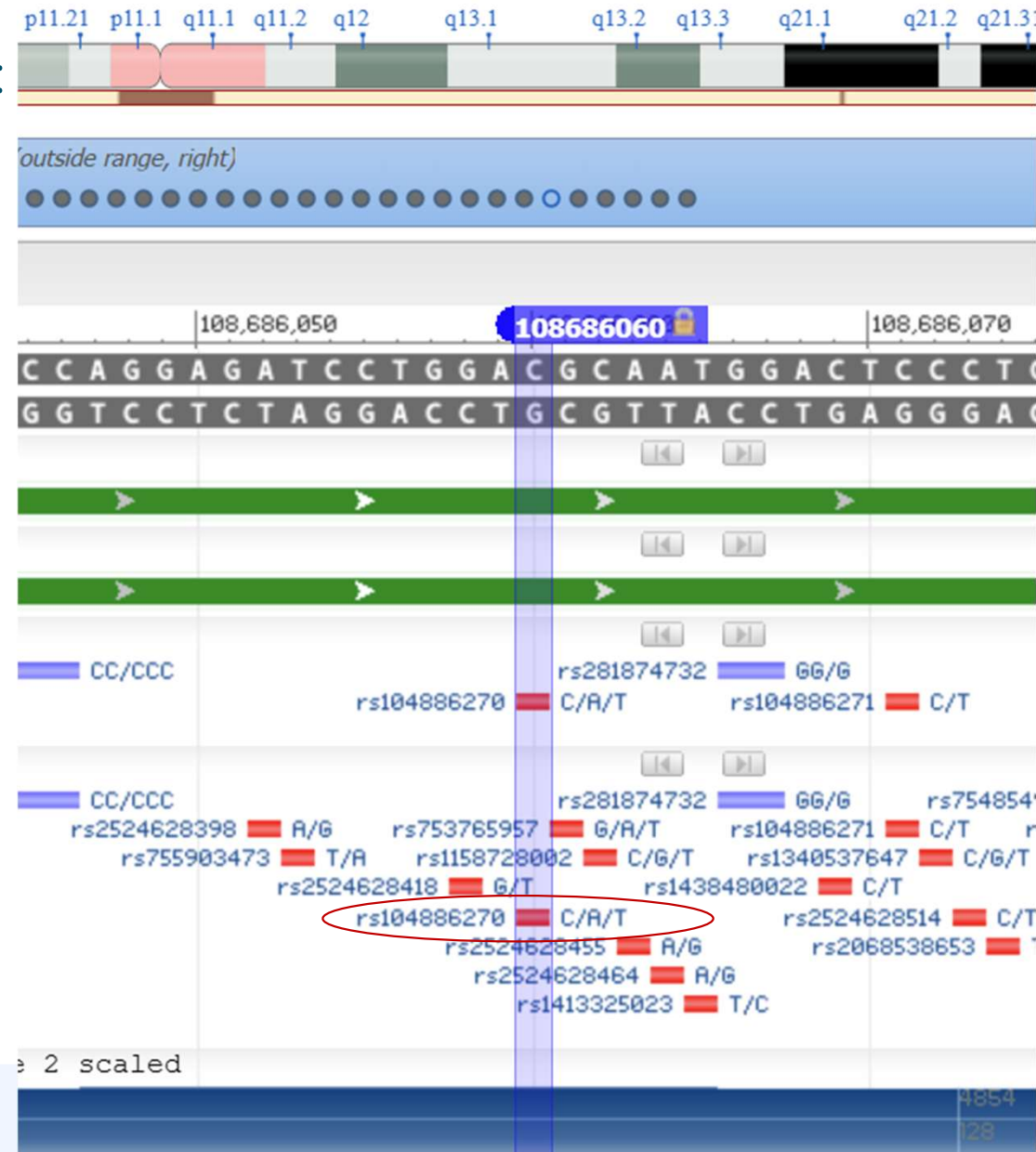
4) Variant interpretation

Categories:

- 5 – pathogenic variant
- 4 – likely pathogenic variant
- 3 – variant of uncertain significance (VUS)
- 2 – likely benign variant
- 1 – benign variant

Read more about the variant at

<https://www.ncbi.nlm.nih.gov/snp/rs104886270>



Help us continue to improve gnomAD by taking 5 minutes to fill out our [user survey](#).

SNV: X-108686060-C-T(GRCh38)

[Copy variant ID](#)[Gene page](#)Dataset [gnomAD v3.1.2](#) ⓘ

Filters ⓘ	Genomes
Allele Count	Pass
Allele Number	9
Allele Frequency	111494
Grpmax Filtering AF ⓘ (95% confidence)	0.00008072
Number of homozygotes	0.00003698
Number of hemizygotes	0
Mean depth of coverage	2
	24.0

External Resources

- [dbSNP \(rs104886270\)](#)
- [UCSC](#)
- [ClinVar \(24718\)](#)
- [ClinGen Allele Registry \(CA259005\)](#)
- [All of Us](#)

Feedback

[Report an issue with this variant](#)

Genetic Ancestry Group Frequencies ⓘ

[gnomAD](#) [HGDP](#) [1KG](#) [Local Ancestry](#)

Genetic Ancestry Group	Allele Count	Allele Number	Number of Homozygotes	Number of Hemizygotes	Allele Frequency
▶ Remaining individuals	1	1490	0	0	0.0006711
▶ Admixed American	1	10512	0	0	0.00009513
▶ European (non-Finnish)	5	53094	0	1	0.00009417
▶ African/African American	2	30628	0	1	0.00006530
▶ European (Finnish)	0	6029	0	0	0.000
▶ Amish	0	683	0	0	0.000
▶ East Asian	0	3539	0	0	0.000
▶ Middle Eastern	0	236	0	0	0.000

dbSNP

SNP



rs104886270



Search

Create alert Advanced

Help

Display Settings: Summary

Send to:

☐ rs104886270 [Homo sapiens]

1.

Variant type: SNV
Alleles: C>A,T [Show Flanks]
Chromosome: X:108686060 (GRCh38)
X:107929290 (GRCh37)
Canonical SPDI: NC_000023.11:108686059:C:A,NC_000023.11:108686059:C:T
Gene: COL4A5 (Varview)
Functional Consequence: genic_downstream_transcript_variant,coding_sequence_variant,missense_variant
Clinical significance: uncertain-significance,conflicting-interpretations-of-pathogenicity
Validated: by frequency,by alfa,by cluster
MAF: T=0.0 (ALFA)
T=0.000049/4 (ExAC)
T=0.000057/15 (TOPMED)

...more

HGVS: NC_000023.11:g.108686060C>A, NC_000023.11:g.108686060C>T, NC_000023.10:g.107929290C>A,
NC_000023.10:g.107929290C>T, NG_011977.2:g.251137C>A, NG_011977.2:g.251137C>T, NM_000495.5:c.4228C>A,
NM_000495.5:c.4228C>T, NM_000495.4:c.4228C>A, NM_000495.4:c.4228C>T, NM_033380.3:c.4246C>A,
NM_033380.3:c.4246C>T, NM_033380.3:c.4246C>A, NM_033380.3:c.4246C>T, NM_044520040.3:c.4264C>A,
NM_044520040.3:c.4264C>T, NM_044520040.3:c.4264C>A, NM_044520040.3:c.4264C>T

...more

Search details

rs104886270[All Fields]

Search

See more...

Recent activity

Turn Off Clear

rs104886270 (1)

SNP

Anemia, sickle cell - Genes and Disease

APC[gene] AND (clinsig_pathogenic[prop]
OR clinsig_likely_pathog... (2524) ClinVar

MYH7[gene] AND (clinsig_pathogenic[prop]
OR clinsig_likely_pathog... (311) ClinVar

HFE[gene] AND (clinsig_pathogenic[prop]
OR clinsig_likely_pathog... (35) ClinVar

See more...

Workflow at the Lab of Medical Genetics:

5) Diagnosis

The most relevant
candidate variants are
interpreted

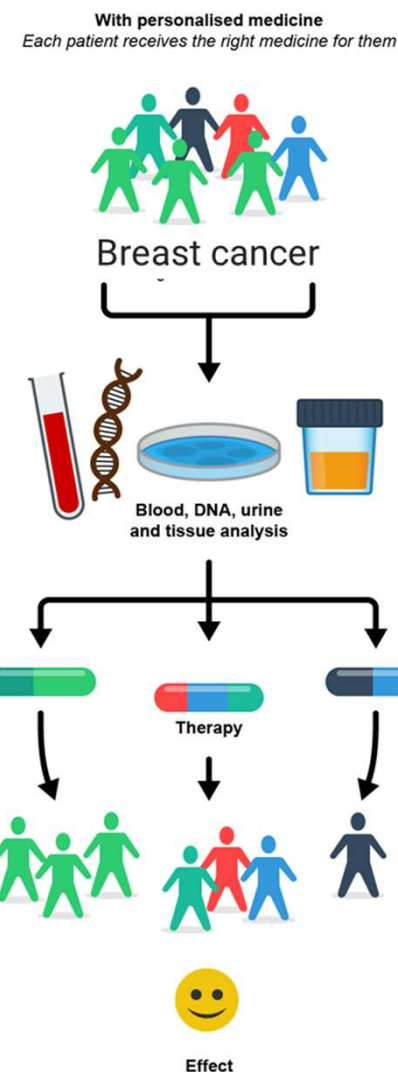
Sometimes a genetic cause
matching the symptoms is
found, in other cases it's not



Personalised Medicine

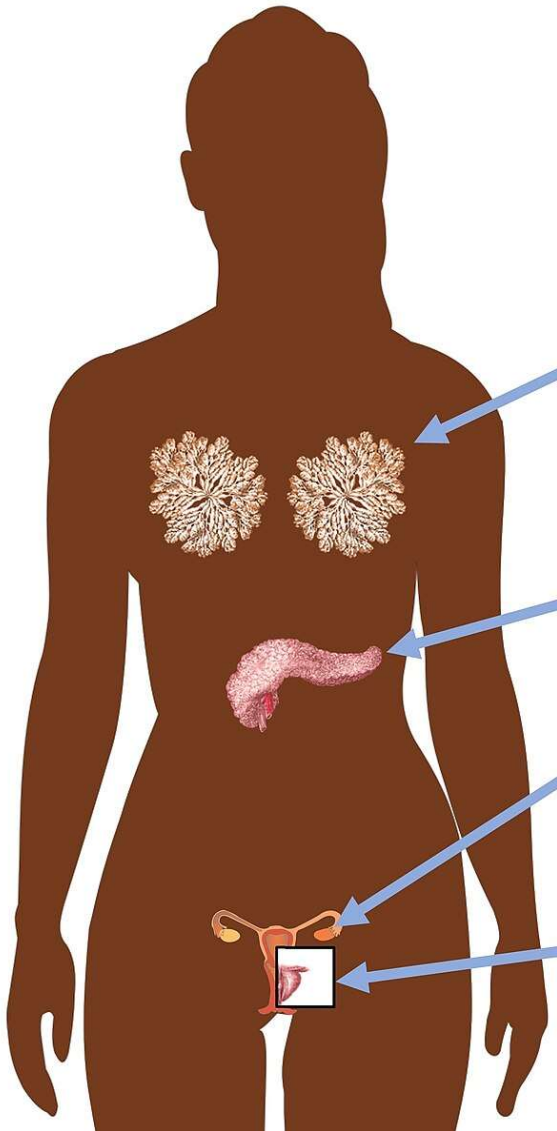


- Diagnosis – fast and correct
- Medical treatment (most common: choose right cancer medicine)

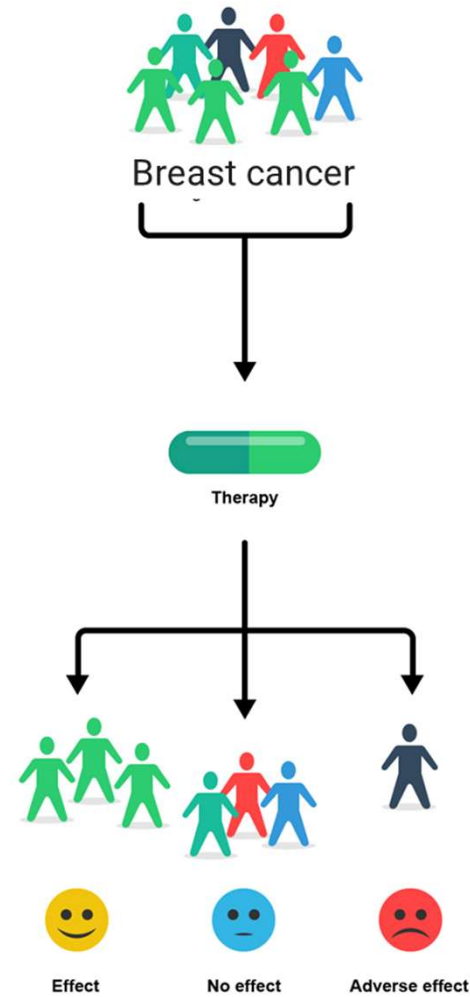


© ABPI 2016

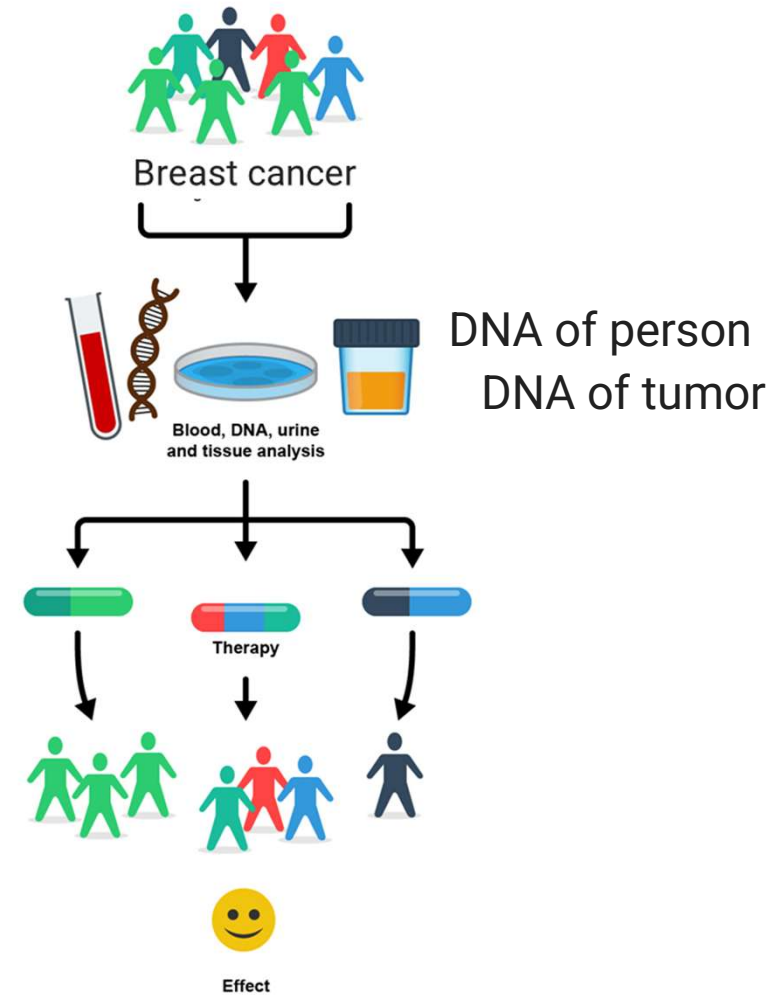
Also see: <https://www.regjeringen.no/contentassets/c0ab0380265445e58508c36e51e5561b/no/pdfs/strategi-for-persontilpasset-medisin.pdf>,
<http://www.helsenorge.no/persontilpasset-medisin>. Image to the right: Example schematic of use of personalized medicine. From [Personalised medicine, ABPI](#)



Without personalised medicine
Some benefit, some do not



With personalised medicine
Each patient receives the right medicine for them



© ABPI 2016

Today's lecture

1. Introduction to Genetics

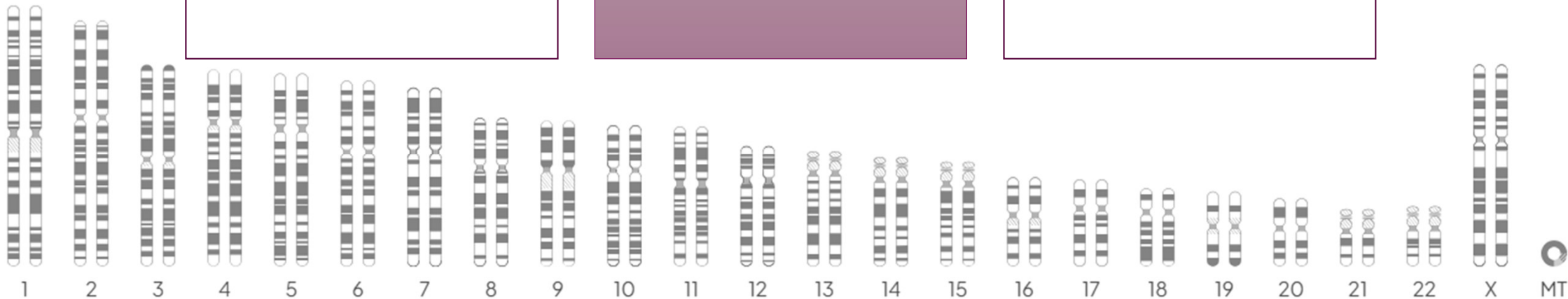
2. Gene Expression and Errors

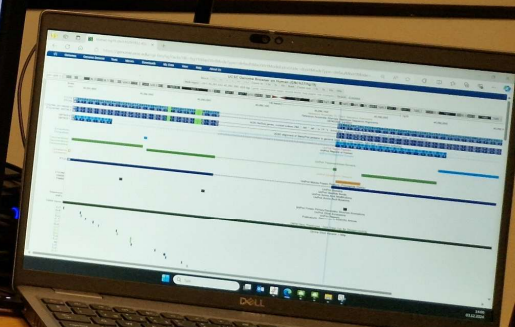
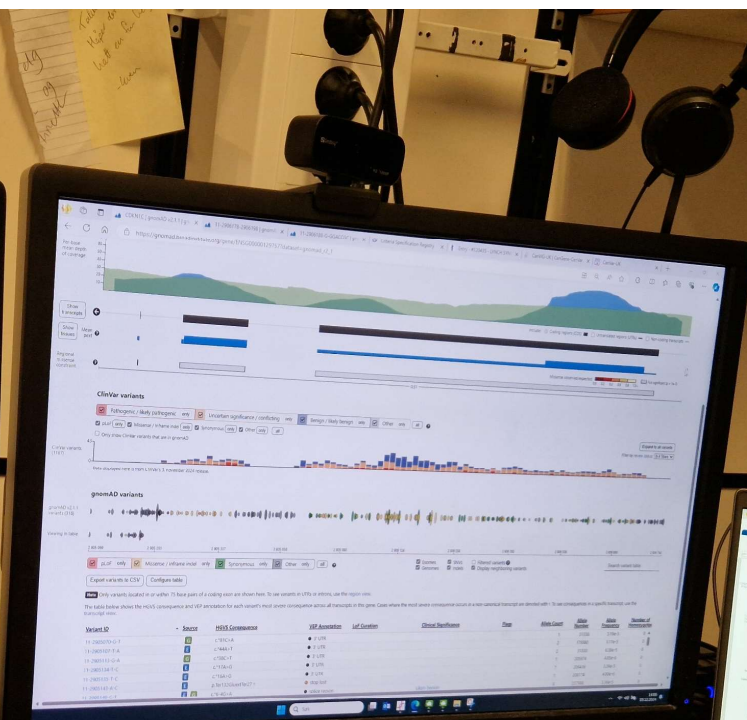
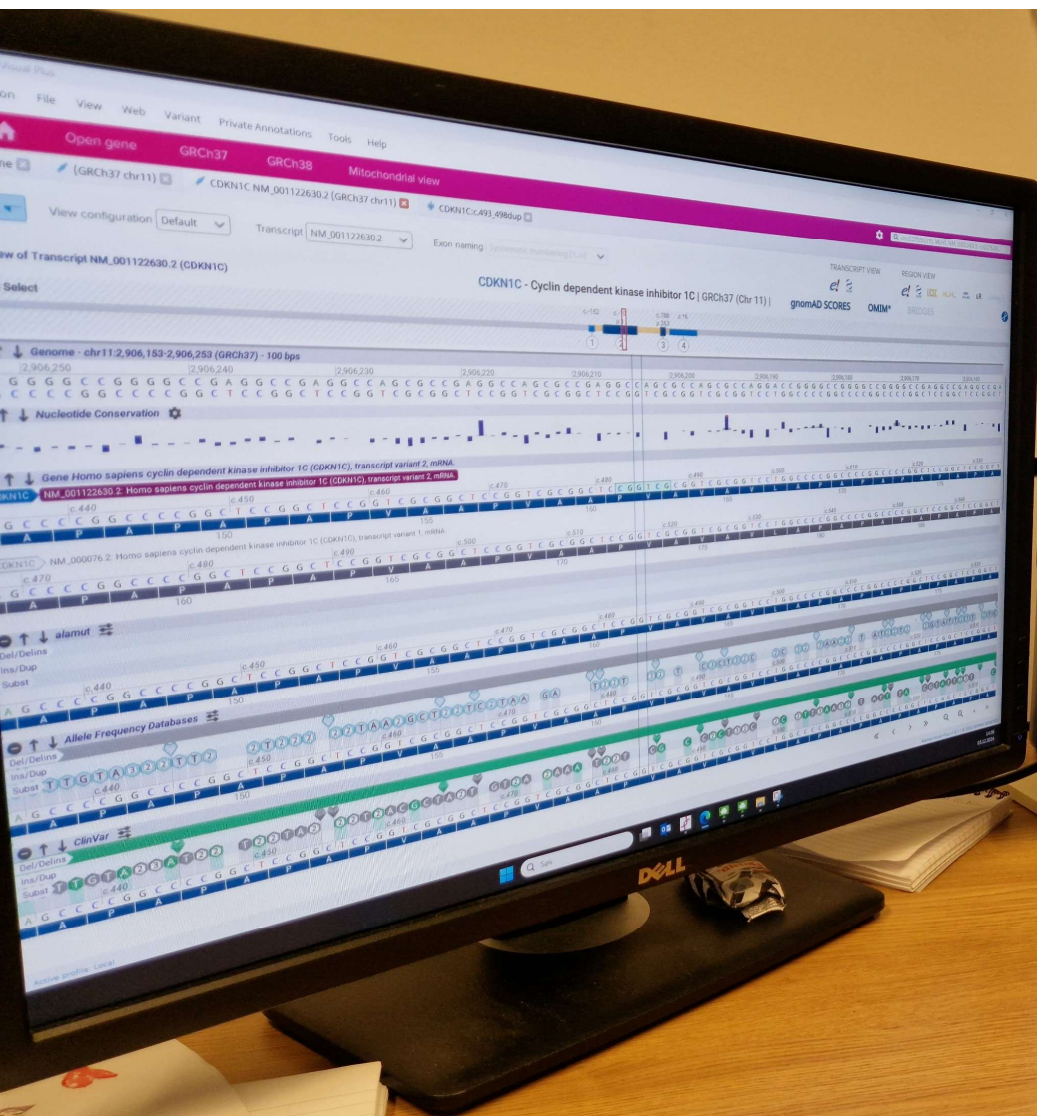
3. Genetic Errors and Conditions

4. Medical Genetics

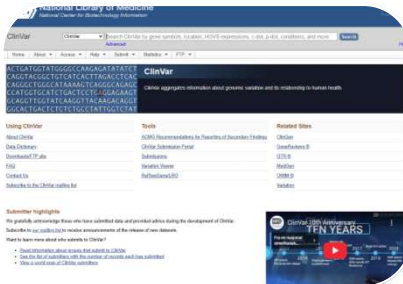
5. IT, Infrastructure and Cooperation

6. Ethical Considerations





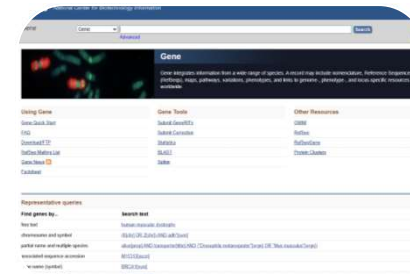
International Databases



ClinVar



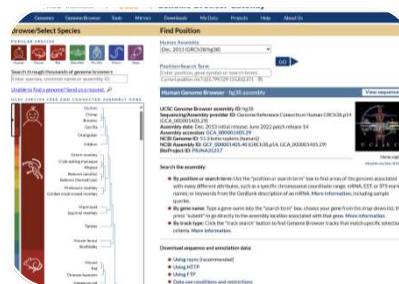
Ensembl



NCBI Gene



RefSeqGene



UCSC Genome Browser



HGMD (need account)

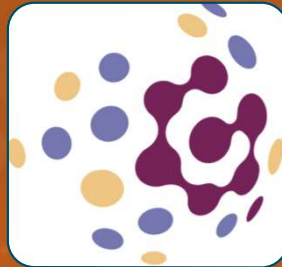
Some important

European Cooperation Initiatives



1+ Million Genomes (1+MG) Initiative

Including the project Genome of Europe



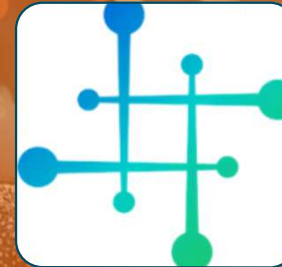
European Genomic Data Infrastructure (GDI)



Horizon Europe



ELIXIR
(European Life-Science Infrastructure for Biological Information)



European Health Data Space (EHDS)



EATRIS
(European Infrastructure for Translational Medicine)

Ongoing Work

Data standards
for genomic
data

Secure
infrastructure
for data sharing

Federated data
access models

Platforms for
secure genomic
data sharing

AI and high-
performance
computing

Genomic
research

Today's lecture

1. Introduction to Genetics

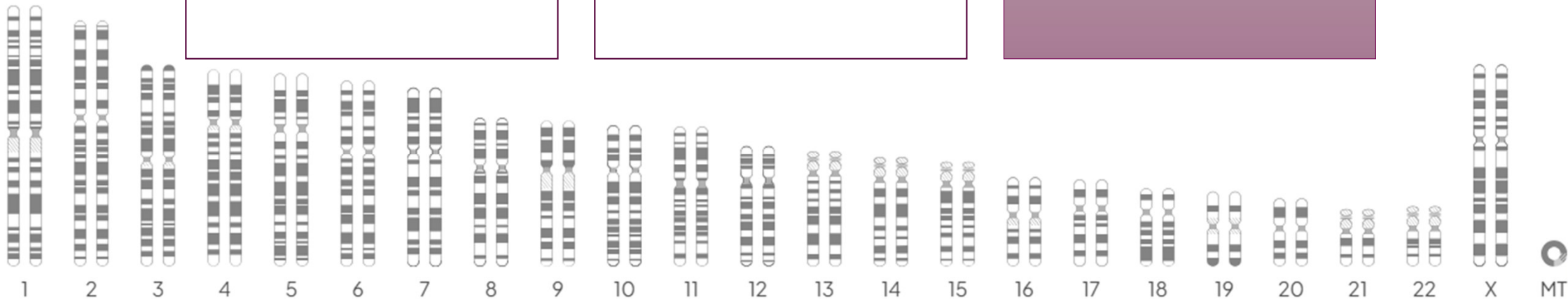
2. Gene Expression and Errors

3. Genetic Errors and Conditions

4. Medical Genetics

5. IT, Infrastructure and Cooperation

6. Ethical Considerations



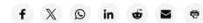
Health

How CRISPR therapy could cure everything from cancer to infertility

The imminent approval of the world's first CRISPR treatment for sickle cell disease is just the start: soon this gene-editing tool could be used to tackle everything from cancer to high cholesterol and infertility

By Michael Le Page

3 July 2023



Neil Webb

The Brussels

TUM

BUSINESS

ART & CULTURE

EU

Dutch scientists cult synthetic human twi

Wednesday 10 April 2024

By The Brussels Times with Bel



Credit: Canva

Scientists at Maastricht University's MERLN institute and Maastricht UMC+ hospital have successfully grown an identical twin human embryonic structure using only stem cells, without the need for eggs or sperm, according to a university press release on Wednesday.



18 February 2025

Artificial human DNA study raises urgent ethical questions for society

Written by: Joe Stafford

Researchers from The University of Manchester - in collaboration with the Universities of Granada, Lund, and Oslo - have conducted the first-ever ethical analysis of artificial human DNA.

The study, published in the *Journal of Medical Ethics*, explores how this emerging technology could impact identity, privacy and even reproduction, raising urgent questions for society.

Artificial human DNA, or synthetic DNA (synDNA), is created by assembling the chemical components of genes in a controlled process. While this technique could revolutionise medicine and biology, it also presents ethical challenges that must be addressed before the technology becomes widespread.

Scientists have already synthesised bacterial genomes and portions of human chromosomes, and future advancements could make it possible to generate entire human genomes in a lab.

Nanobots Will Be Flowing Through Your Body by 2030

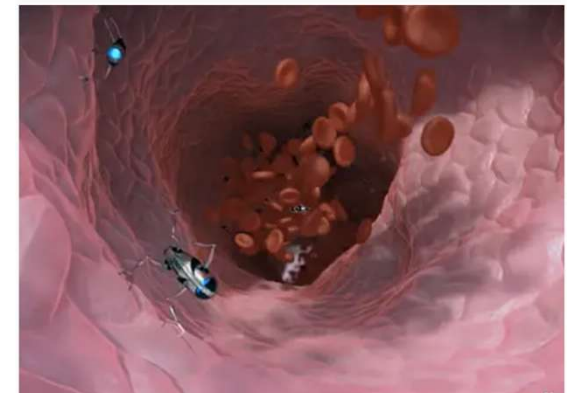
In 10 years, nanobots in your blood might keep you from getting sick, transmit your thoughts to a wireless cloud, and give you eternal life.

Updated: Apr 03, 2023 02:42 AM EST



Trevor English

4 years ago



Dimension Studios/YouTube

Futurists often claim that nanobots hold the key to eternal life. Inventor, author, and Google futurist [Ray Kurzweil](#), in an interview with [Neil deGrasse](#), claimed that tiny nanobots in our bloodstreams will prevent us from dying in 2030. According to Kurzweil, the nanobots will travel inside you on a molecular level, protecting your biological system and ensuring you'll have a [good and long life](#). The future is closer than you may think!

Nanobots injected into your bloodstream

IFL Science claims that [DNA robots](#) are already being tested in animals to seek out and destroy cancer cells. These programmed strands of DNA can move through the

Read more: <https://biotechnologyjobs.co.uk/industry-insights/biotechnology-sector-predictions-for-the-next-5-years-technological-progress-emerging-applications-and-the-evolving-job-market>, [Nanobots Will Be Flowing Through Your Body by 2030](#)

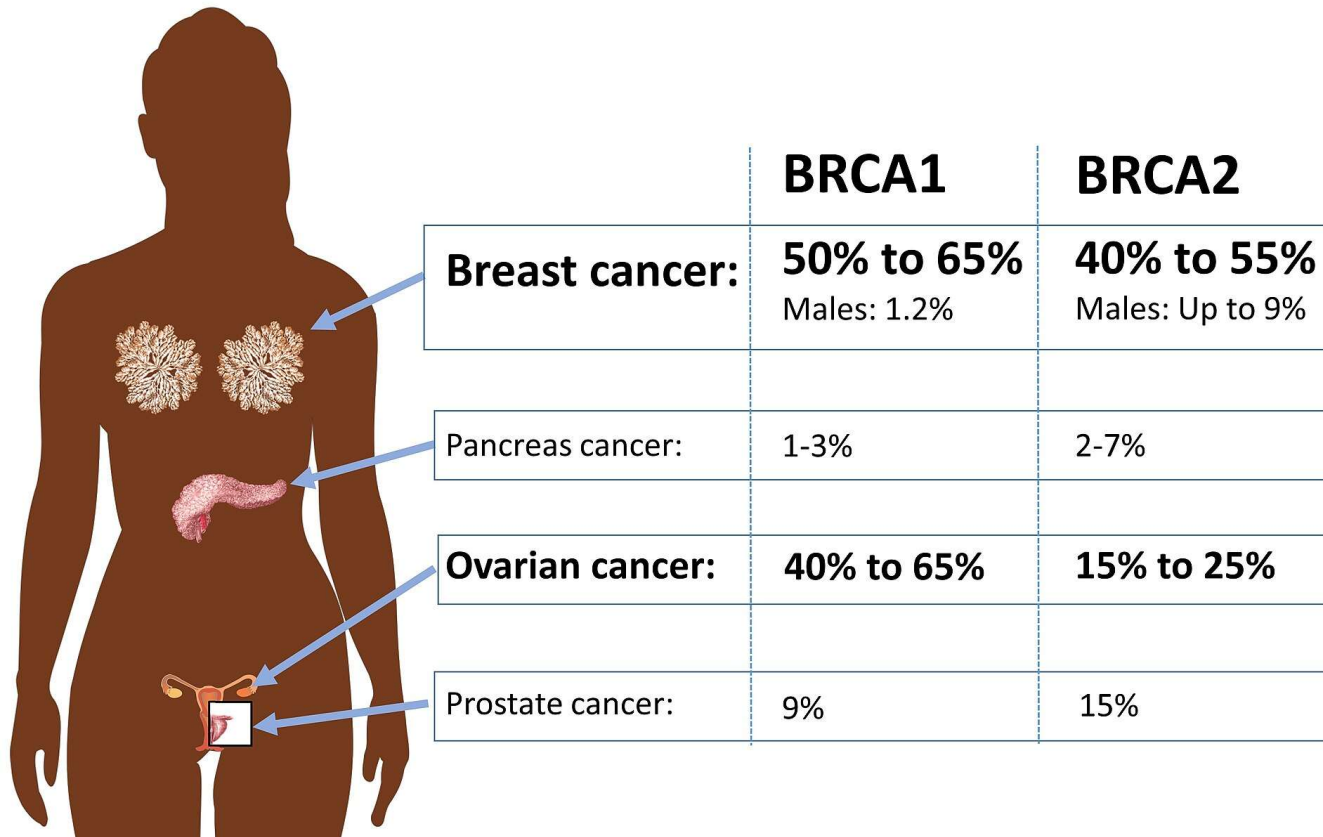
Ethical Dilemmas

What contributes (most) to a good life?

Common values and principles in medical ethics:

- Autonomy (including informed consent)
- The right to know – and the right not to know (about the future)
- Do no harm
- Promote well-being
- Justice
- Trust
- ... and more





In a family with multiple cases of breast cancer, several members carry a disease-related gene variant.

A man, whose mother died of breast cancer, is advised to get genetic counseling and testing, but he declines because his own risk is low.

However, his adult daughter could be at high risk if she inherited the variant.

The doctor faces a conflict between 1) respecting the man's autonomy and 2) protecting the daughter's right to know, while confidentiality prevents disclosing the man's genetic information without his consent.

Petrucci N, Daly MB, Pal T (September 1998). "[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)". In Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJ, Gripp KW, Amemiya A, editors (eds.). *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle. [PMID 20301425](#).

Work together in groups with an ethical dilemma

Exercise:

Choose one of the following ethical dilemmas, and discuss it in groups

You may use the SME model if you want to

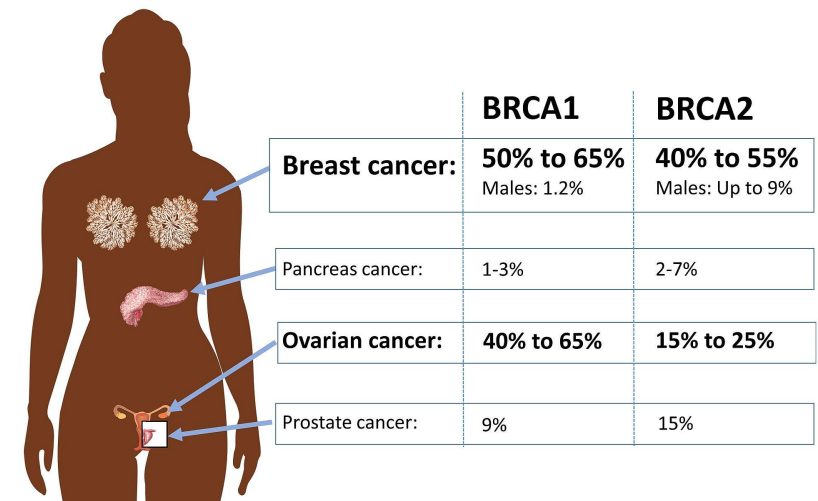
The SME-model

1. Define the ethical problem(s)
2. Describe facts
3. Provide details of the parties involved
4. Identify the values, ethical principles and laws at stake
5. Based on the above, identify and discuss possible solutions of the case
6. Conclusion, follow up and evaluation

<https://www.med.uio.no/helsam/tjenester/kunnskap/etikk-helsetjenesten/sme-modellen/index.html>

Dilemma: Do you want to know about genetic findings?

- If you agreed to participate in research based on your DNA, would you want to be informed if you carried genetic variants associated with diseases that you are highly likely to develop, and for which treatment is available—and why or why not?
- Examples of such genetic variants:
 - Variants that increase the risk of cancer like the BRCA1 and BRCA2 (see the picture to the right)
 - Variants associated with heart conditions that can lead to sudden death as the first symptom



What considerations would be important to you when deciding whether to receive results about such genetic findings?

Vil tillate gentesting av idrettsutøvarar



Av: Svein Isungset Støve
Publisert: 19. februar 2026

For å kunne delta i kvinneklassa i VM i friidrett og boksing hausten 2025, måtte norske landslagsutøvarar ta ein gentest som er ulovleg å gjera i Noreg. No ønsker fleirtalet i Bioteknologirådet å endre forbodet.

Det er OL i Rio i 2016. Startskotet nærmar seg i finalen på 800 meter for kvinner. På startstreken står åtte av verdas beste løparar, klare for å springa sitt livs løp. Blant dei var sør-afrikanske Caster Semenya, som ufrivillig har vorte ein av dei mest kontroversielle deltakarane i kvinneklassa i friidrett på 2010-talet. Semenya har kvinnelege kjønnskarakteristikkar, men er fødd med mannlege kjønnskromosom og ein genetisk variant som påverkar korleis kroppen responderer på testosteron. Dette vert kalla variasjon i kroppslig kjønnsutvikling (VKK), også ofte kalla DSD (sjå faktaboks). I finalen i Rio la Semenya seg tidleg i front, og vann til slutt gull. Sølvet gjekk til Francine Niyonsaba frå Burundi og bronzen til Margaret Wambui frå Kenya. Alle dei tre utøvarane har mannlege kjønnskromosom og ein DSD-tilstand. Resultatet frå denne OL-finalen sette fart på diskusjonar kring kor vidt desse utøvarane har ein urettferdig fordel i konkurranse med personar fødd med kvinnelege kjønnskromosom, og kring kven som burde, og kven som ikkje burde, få lov til å delta i kvinneklassa.

SRY-genet, kjønshormon og kjønntesting

SRY er eit gen som ligg på Y-kromosomet og er nødvendig for å starte utvikling av testiklar i fosterlivet. Testiklar produserer hormona testosteron og AMH, som påverkar vidare kjønnsutvikling.

Testosteron har innverking på muskelmasse, styrke og oksygentransport.

Dilemma: Sex Verification Testing of athletes

- SRY is a gene located on the Y chromosome that is necessary for initiating male development, including traits such as greater muscle mass, strength, and oxygen transport.
- Some sports organizations are considering introducing genetic testing for the SRY gene
- The test is carried out to ensure that women in the women's category compete under equal conditions
- Women with the SRY gene will be excluded from competitions. Some of these women may not be aware that they carry the SRY gene before testing

Define different perspectives and ethical challenges

Dilemma: DNA test reveals he is not the child's father

A child has a serious illness that doctors haven't been able to diagnose.

They perform a so-called trio test, which is a genetic test of both parents and the child, to identify de novo mutations in the child (mutations that are not present in the healthy parents).

The test fail, since it reveals that the man is not the child's biological father.

Define ethical challenges

If you were a staff member at this laboratory, what would you do? List alternatives with advantages and disadvantages

Når DNA-test viser at pappa ikke er far

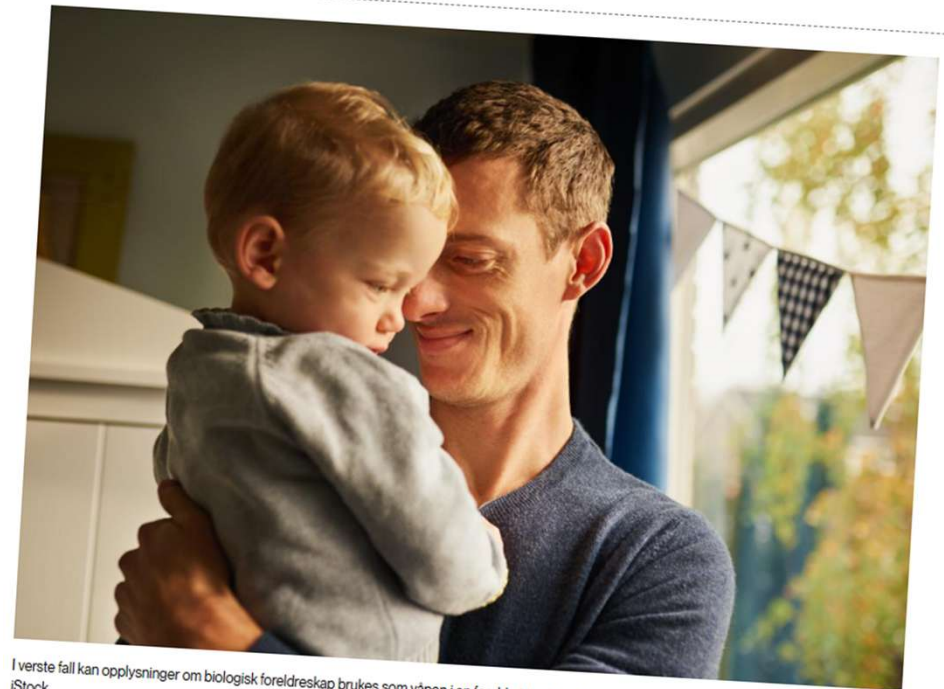
– Slik lovverket er i dag, finnes det ingen grense for hvor lenge foreldre kan tilbakeholde opplysninger om biologisk

Simon Gramvik

Forskningsjournalist

simon@kilden.forskningsradet.no

29. juni 2023



I verste fall kan opplysninger om biologisk foreldreskap brukes som våpen i en foreldretvist, forteller jusprofessor Kirsten Sandberg. Illustrasjonsfoto: iStock

Sprek | Gentesting

Legen tok DNA-test: – Føltes som jeg fikk en sluttdato for livet mitt.

Hun har møtt pasienter i krise og ofte møtt døden. Men da legen Anna Grönlund fikk resultatet av sin egen DNA-test, ble hun overrasket over egen reaksjon: – Jeg tok det enormt hardt.

[▶ Lytt til artikkelen • 7 min](#)

Anna Grönlund har gjort store endringer i livet sitt etter DNA-beskjeden. Foto: Karl Melander

Viktoria Myrén
Svenska Dagbladet

Publisert: 11.01.2025 12:02

Dilemma: Do you want to live with or without the knowledge of a serious future diagnosis?

Discuss arguments for and against being informed about the risk of diseases that cannot currently be cured.

Text from the article:

"I had the idea that I would be able to interpret the results myself, despite knowing that it involved an enormous amount of raw data", she says, shaking her head at her own naïveté

With 95 percent certainty, she will have measurable amyloid plaques in her brain—a hallmark of Alzheimer's—by the age of 65. Whether this will then affect her cognitive abilities, she does not know. But it is likely.

Dilemma: Is it acceptable for commercial companies to sell gene databases?

Discuss arguments for and against.



UNIK MULIGHET: Norge har en unik mulighet til å være en global leder innen etisk og ansvarlig helseteknologi, mener Ira Haraldsen. Foto: Tone Herregården

Når genene våre blir «big business»

Når genene våre legges ut på det frie markedet som dagens fangst, til høystbydende, så er det desto viktigere å holde hodet kaldt.

Ira Haraldsen
PROSJEKTLEDER, AIMIND

PUBLISERT 02.01.2025 - 08:46

Gentesting: Hvor mye vil du egentlig vite?

Det finnes flere private tilbydere av genetiske selvtester som sier noe om din genetiske disposisjon for sykdommer. Men vil resultatet gjøre livet ditt bedre?



DNA: Gentester kan fortelle om dine arvelige egenskaper og sykdomsrisikoer. Det er arvestoffet (DNA) som undersøkes. Foto: NTB Scanpix / Shutterstock

Dilemma: When you wish to be informed about a genetic condition, while your relatives prefer not to know.

Is it acceptable for you to take a genetic test to find out if you have inherited a disease gene, when your parents do not want to know whether they have inherited it?

Discuss arguments for and against undergoing genetic testing

Dilemma: Healthy child?

Some fertility companies offer to compare the mother's and donor's genes to avoid shared disease genes and increase the chance of a healthy child.

Would you use this service if it were up to you?

Why – why not? Argue

The screenshot shows the website for the European Sperm Bank, with the tagline "— becoming Ferti". At the top right is a red button labeled "Find sperm donors" and a search icon. The main heading is "Your genes carry genetic disease". Below it, the text explains that everyone is a carrier of genetic disease due to mutations, which can cause serious disease in children. It states that a child inherits a genetic disease when both parents have mutations in the same gene. To reduce this risk, the company has developed GeneXmatch. A central graphic displays ten vertical bars, each representing a pair of chromosomes with red and blue bands. Below this, a white box titled "What is GeneXmatch?" describes the screening test. It explains that the test compares the prospective mother's genes with her chosen sperm donor's genes to determine if they share mutations in the same genes, which could put the future child at risk of a serious disease. It notes that GeneXmatch screens for over 400 genetic diseases, providing an added sense of security. A link "See what we screen for here" is provided at the bottom of the box. On the right side of the page, there is a photograph of a baby with blue eyes, holding a yellow and red object in its mouth.

EUROPEAN
SPERM-BANK
— becoming Ferti

Find sperm donors

Your genes carry genetic disease

We're all carriers of genetic disease. That's because our genes contain errors - or mutations. Even if we're not sick ourselves, our mutations can cause serious disease in our children.

In many cases, a child inherits a genetic disease when the man and the woman have mutations in the same gene.

To reduce the risk of having a child with genetic disease, we've developed GeneXmatch.

What is GeneXmatch?

GeneXmatch is a screening test that improves the chances of your future baby being healthy.

The test compares the genes of the prospective mother with her chosen sperm donor. By matching their genes, we're able to determine if the woman and her donor carry mutations in the same genes. In other words, we can tell if a future child is at risk of inheriting a serious disease.

GeneXmatch screens for more than 400 genetic diseases. That way, you get an added sense of security that you've chosen the right donor.

[See what we screen for here](#)

Regelverk, juridisk rettleiing og avklaringar

Få oversikt over relevant regelverk og veiledere, og få veiledning.

Prosess og hjelp til å løyse juridiske problemstillinger

Tverretatleg rettleiingsteneste

Svar på spørsmål i oppdragsbrev fra Helse Sør-Øst RHF

[PDF](#) →

Helseforskningslovens virkeområde - godkjenning for biobank og innførsel av biologi (2014)

[PDF](#) →

Spørsmål om forholdet mellom lov om behandlingsbiobanker og helseforskningsloven (2014)

[PDF](#) →

Svar på henvendelse om helseforskningslovens anvendelsesområde (2021)

[PDF](#) →

Vedrørende spørsmål om lovverk som regulerer helseforskning og bruk av anonymisert (2014)

[PDF](#) →

Read more here: <https://www.helsedirektoratet.no/forebygging-diagnose-og-behandling/diagnose-og-behandling/persontilpasset-medisin/regelverk>

Søk

Meny

HELSE

norge

Meny

Søk

Forsiden

Forebygging, diagnose og behandling

Diagnose og behandling

< Forside

Strategi for persontilpassa medisin 2023–2030

Regjeringa sin visjon er at persontilpassa medisin skal vere ein integrert del av forebygging, diagnostikk, behandling og oppfølging i helsetenesta, med mål om betre helse og meistring gjennom livet.

Tverretatleg rettleiingsteneste

Få tverretatleg ein-til-ein-retteiing etter fleire regelverk samstundes.

Fagråd for persontilpassa medisin

Fagråd for persontilpassa medisin skal sikre samordna og koordinert oppfølging og implementering av persontilpassa medisin i helsetenesta. Helsedirektoratet leier fagrådet.

Juridiske og etiske vurderingar av Nasjonalt genomsenter

Helsedirektoratet har vurdert etiske og juridiske sider ved etablering av eit nasjonalt genomsenter med n

Rettslege avklaringar om persontilpassa medisin

Uthvalde rettslege avklaringar om persontilpassa medisin

Bioteknologirådet

in

UK

Søk

Meny

Nyheter

Temasider

For skoler

Arrangementer

GENialt

Biotekpodden

Uttalelser

For presse

Sp

Rapporter

Juridiske og etiske s

Forslag til endringer

«Vi gir råd om etisk bruk av bioteknologi på mennesker, dyr, planter og mikroorganismer.»

Aktuelt:

GENialt

SmåGENialt: Jakt på de gode genene

CRISPR

13 år med forskning på Crispr-laks

Epigenetikk

Uenighet om aldring

Persontilpasset medisin

Persontilpasset medisin betyr en skreddersydd behandling som er tilpasset din genetiske profil. For å kunne gi deg en persontilpasset behandling, er det nødvendig å vite mest mulig om dine biologiske forhold. Da vil du kunne få riktig behandling til rett tid.

HELSE

norge

Meny

Søk

< **Medisiner og medisinbruk**

Analyse av gener for å finne riktig medisin (Farmakogenetisk analyse)

Gene dine kan påvirke hvordan du reagerer på medisiner. En gentest (farmakogenetisk analyse) kan i mange tilfeller hjelpe legen din til å velge riktig medisin og dose til deg.

Innhold på siden

- [Hva er farmakogenetisk analyse?](#)
- [Når er farmakogenetisk analyse aktuelt?](#)
- [Hvordan gjøres undersøkelsen?](#)
- [Hvor lagres resultatene av farmakogenetiske analyser?](#)

Siv Anette Fjellkårstad

Siv.Anette.Fjellkarstad@helsedir.no

Logg inn

NorPreM

Nasjonalt kompetansenettverk for persontilpasset medisin

Om nettverket

Prosjektportefølje

Har du en prosjektidé?

Arrangementer

Internasjonale konferanser

Regionale NorPreM-råd

Film/intervjuer

Resultater og rapporter

3. februar 2025

Nytt prosjekt i NorPreM – AmyPrem

13. oktober 2024

Velkommen til fagdag i PM i Helse Midt – Norge

10. september 2024

Hvordan gikk det når Juss møtte medisin?

10. september 2024

Nye videoer fra NorPreM arrangementer

Search (hit return for all results)

Søk

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GeNotes

GTAC

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News & Events

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NHS England Genomics Education Programme

Delivering genomics education, training and experience for the healthcare workforce

Learn about the impact of genomics

Try our educational opportunities for health professionals. Get started below, or browse our full collection.

FEATURED PROFESSION

BITESIZE LEARNING

SHORT COURSE

Thank you!

1. Introduction to Genetics

2. Gene Expression and Errors

3. Genetic Errors and Conditions

4. Medical Genetics

5. IT, Infrastructure and Cooperation

6. Ethical and Legal Considerations

